

**What are the Adult Outcomes after
Anorectal Malformation Repair?**

By

**Melissa Clare Davies
MB BS, MRCS**

**A thesis submitted to the University of London for the
degree of Doctor of Medicine**

July 2007

UMI Number: U593167

All rights reserved

INFORMATION TO ALL USERS

The quality of this reproduction is dependent upon the quality of the copy submitted.

In the unlikely event that the author did not send a complete manuscript and there are missing pages, these will be noted. Also, if material had to be removed, a note will indicate the deletion.



UMI U593167

Published by ProQuest LLC 2013. Copyright in the Dissertation held by the Author.
Microform Edition © ProQuest LLC.

All rights reserved. This work is protected against
unauthorized copying under Title 17, United States Code.



ProQuest LLC
789 East Eisenhower Parkway
P.O. Box 1346
Ann Arbor, MI 48106-1346

STATEMENT OF ORIGINALITY	10
ACKNOWLEDGMENTS	11
PRESENTATIONS TO LEARNED SOCIETIES	12
CHAPTER 1- INTRODUCTION	14
INTRODUCTION	14
BACKGROUND TO RESEARCH QUESTION	14
EMBRYOLOGY	16
AETIOLOGY AND EPIDEMIOLOGY	20
CLASSIFICATION	22
EXISTING LONG-TERM OUTCOME DATA	23
<i>Urinary Function</i>	23
<i>Bowel Function</i>	24
<i>Gynaecological Outcomes</i>	27
<i>Male Outcomes</i>	29
<i>Psychological Outcomes</i>	30
CONCLUSIONS	31
CHAPTER 2 - TREATMENT OF ANORECTAL MALFORMATIONS	32
INTRODUCTION	32
HISTORICAL CONSIDERATIONS	32
CURRENT MANAGEMENT	33
<i>Primary Surgery</i>	34
<i>Secondary Surgery</i>	37
<i>Urinary</i>	42
<i>Genital</i>	44
CONCLUSIONS	45
CHAPTER 3 – METHODS & TECHNIQUES	46
INTRODUCTION	46
TRACING & RECRUITMENT	48
QUESTIONNAIRES	48
<i>Bowel Function</i>	49
<i>Lower Urinary Tract Symptoms Questionnaire</i>	50
<i>Quality of life</i>	51
<i>Sexual Function</i>	52
<i>Body Esteem</i>	54
DISCUSSION	55
<i>Bowel Function</i>	56
<i>Quality of life</i>	56
<i>Sexual Function</i>	56
<i>Body Esteem</i>	57
CONCLUSIONS	57
CHAPTER 4 - GENERAL RESULTS	58
INTRODUCTION	58
DEMOGRAPHICS	60
CLINICAL DATA	61
<i>ARM classification</i>	61
<i>Associated Anomalies</i>	63

DISCUSSION.....	64
<i>Demographics</i>	64
<i>Clinical Data</i>	65
CONCLUSIONS	65
CHAPTER 5 - BOWEL FUNCTION	67
INTRODUCTION.....	67
RESULTS.....	67
<i>Mode of Defecation</i>	67
<i>Cleveland Clinic Incontinence Score - CCIS</i>	68
<i>CCIS Scores in patients requiring assistance</i>	69
<i>Associated anomalies</i>	70
<i>Type of repair</i>	70
<i>Constipation Scoring System</i>	71
<i>Quality of Life</i>	72
DISCUSSION.....	73
<i>Incontinence</i>	73
<i>Constipation</i>	74
<i>Type of Initial Repair</i>	75
<i>ACE</i>	75
CONCLUSIONS	76
CHAPTER 6 - URINARY FUNCTION	78
INTRODUCTION.....	78
RESULTS.....	79
<i>GU Anomalies</i>	79
<i>Mode of Voiding</i>	80
<i>Incontinence</i>	81
<i>Lower Urinary Tract Symptoms (LUTS)</i>	82
DISCUSSION.....	84
<i>Associated Anomalies</i>	84
<i>Incontinence</i>	84
<i>LUTS</i>	85
<i>Neuropathic Bladder</i>	86
CONCLUSIONS	87
CHAPTER 7 –SEXUAL FUNCTION & FERTILITY	88
INTRODUCTION.....	88
RESULTS.....	89
<i>Fertility</i>	89
<i>Genital Anomalies</i>	90
DISCUSSION.....	96
<i>Genital anomalies</i>	97
<i>Sexual Satisfaction</i>	98
CONCLUSIONS	101
CHAPTER 8- PSYCHOLOGICAL OUTCOMES.....	102
INTRODUCTION.....	102
PATIENTS AND METHODS	103
RESULTS.....	104
<i>Body Esteem</i>	104
<i>Quality of Life</i>	106
<i>Impact of condition on ability to form relationships</i>	109
DISCUSSION.....	109
<i>Body Esteem</i>	110
<i>Quality of Life</i>	111
<i>Impact of condition on ability to form relationships</i>	112
CONCLUSIONS	112
CHAPTER 9 – CONCLUSIONS	113

<i>Bowel Function</i>	113
<i>Urinary Function</i>	114
<i>Sexual Function & Fertility</i>	115
<i>Psychological Function</i>	116
RECOMMENDATIONS FOR FUTURE WORK	117
BIBLIOGRAPHY	119
APPENDIX 1	136
APPENDIX 2	137
APPENDIX 3	139
APPENDIX 4	162

Table of Tables

TABLE 1 – PENA CLASSIFICATION OF ANORECTAL MALFORMATIONS	22
TABLE 2– SUMMARY OF BOWEL FUNCTION RESULTS (34).	26
TABLE 3– SUMMARY OF PATIENTS UNABLE TO PARTICIPATE.....	58
TABLE 4–SUMMARY OF SOCIOECONOMIC CLASS (SEC)	60
TABLE 5 - SUMMARY OF DIAGNOSIS.....	61
TABLE 6 – SURGICAL CORRECTION OF ARM – INITIAL PROCEDURES	62
TABLE 7 – NUMBERS OF ASSOCIATED ANOMALIES	64
TABLE 8 – SUMMARY OF MODE OF DEFAECATION.....	68
TABLE 9 – SUMMARY OF CLEVELAND CLINIC INCONTINENCE SCORES AND LEVEL OF ARM.....	69
TABLE 10–CLEVELAND CLINIC INCONTINENCE SCORES IN PATIENTS REQUIRING ASSISTANCE WITH DEFECACTION	70
TABLE 11 – SF 36 MEAN SCORES OVER ALL CATEGORIES.....	72
TABLE 12 - SUMMARY OF URINARY TRACT ANOMALIES	79
TABLE 13 – METHODS OF BLADDER EMPTYING.....	80
TABLE 14– SUMMARY OF RATES OF URINARY INCONTINENCE	82
TABLE 15 – FREQUENCY OF LUTS.....	83
TABLE 16 – COMPARISON OF LUTS IN FEMALES WITH ARM, CAH AND NORMAL CONTROLS.....	84
TABLE 17 - SUMMARY OF FEMALE GENITAL ANOMALIES.....	90
TABLE 18 - SUMMARY OF MALE GENITAL ANOMALIES	92
TABLE 19– MSQ RESULTS	94
TABLE 20 – MALE SEXUAL SATISFACTION SCORES	95
TABLE 21– SEXUAL SATISFACTION SCORES IN SUB-GROUPS.	96
TABLE 22 – BODY ESTEEM RESULTS FOR MALES	105
TABLE 23– BODY ESTEEM RESULTS FOR FEMALES.....	106
TABLE 24 - SUMMARY OF SF-36 RESULTS.....	107

Table of Figures

FIGURE 1 – DIVISION OF THE CLOACA AT 4 WEEKS (9)	16
FIGURE 2 - DIVISION OF THE CLOACA AT 6 WEEKS (9)	17
FIGURE 3 - DIVISION OF THE CLOACA AT 7 WEEKS (9)	17
FIGURE 4 –LOW ARM, ANAL STENOSIS AND ANAL ATRESIA (9).....	18
FIGURE 5 – HIGH ARM ANORECTAL MALFORMATION (9)	19
FIGURE 6 - MULLERIAN DUPLICATION AS A RESULT OF CLOACAL ANOMALY (11)	19
FIGURE 7 - POSTERIOR SAGITTAL INCISION (73)	35
FIGURE 8 - SEPARATION OF THE RECTUM FROM THE URETHRA (73)	36
FIGURE 9 - THE RECTUM IS PASSED IN FRONT OF THE LEVATOR MUSCLE (73).....	36
FIGURE 10 –ANOPLASTY (73).....	36
FIGURE 11 – ANTEGRADE CONTINENCE ENEMA PROCEDURE (88)	39
FIGURE 12 - GRACILIS MUSCLE TRANSPLANTATION (95).	41
FIGURE 13 – AGE DISTRIBUTION OF PARTICIPANTS	59
FIGURE 14 - ILEAL VAGINA	91
FIGURE 15 – PHOTOGRAPH OF A PATIENT WITH A CLOACA ANOMALY	100
FIGURE 16 – SCATTER PLOT OF SEXUAL ATTRACTIVENESS AND GENERAL HEALTH	108

“To build may have to be the slow and laborious task of years. To destroy
can be the thoughtless act of a single day”

Sir Winston Churchill

(1874 - 1965)

ABSTRACT

Introduction & Objective

Anorectal malformations are a spectrum of anomalies requiring surgery within the first days of life. Many patients have associated anomalies of the genitourinary tract, heart and spine. They may have a significant level of morbidity even after surgical repair. This study looks at the range and degree of morbidity that this group of patients experience.

Methods

A cohort study was performed on adult patients treated at a tertiary referral paediatric hospital as children. Patients were invited to participate in the study; those who agreed were sent questionnaires to complete; consisting of validated questionnaires on urinary, bowel, sexual, quality of life, body image and fertility outcomes (paternity in males). On return of these questionnaires, patients were invited to attend clinic for a physical examination.

Results

117 patients were contacted of whom 75 agreed to take part in the study (64%). The age range was 18-60 years (mean 26 years). 44 of the respondents were females and 31 were male.

57 passed urine without any assistance, 9 required intermittent self-catheterisation (ISC), and 7 had an appendico-vesicostomy, 1 had an ideal conduit and 1 patient expressed her bladder manually. Urinary incontinence was seen in 31 out of 75 respondents (41%).

44 out of the 75 (59%) were able to defecate without any assistance, the rest required assistance of some sort. 11 had an antegrade continence enema, 6 performed regular rectal washouts, 7 had permanent stomas, and 7 used medication to manipulate their bowel movements. Using the Cleveland clinic incontinence score 38 (51%) had either perfect or good continence, 29 (39%) had moderate to severe faecal

incontinence. 8 of the 75 did not answer questions regarding their bowel function as they had permanent stomas.

45 of the 75 (60%) were in a relationship of whom 30 were cohabiting. 27 recorded pregnancies resulted in 20 live births, half of these were to the partners of the male participants. 66 were in full time employment or education with only 9 (12%) registered as disabled.

Conclusions

This study confirms that a significant level of morbidity exists within this patient group. Levels of urinary and faecal incontinence were high. Despite this the majority were in full time employment or education and 60% were in relationships.

STATEMENT OF ORIGINALITY

The composition of this thesis has been entirely my work. It has not been previously submitted.

All clinical data were obtained between October 2003 and September 2005 whilst I was employed by University College London. Honorary contracts were held with Great Ormond Street Hospital and University College Hospitals during the same period.

I certify that I have seen all of the referenced articles quoted.

ACKNOWLEDGMENTS

I would like to thank a number of people for their help and advice during my time in research that made writing this thesis possible.

I am very grateful to my supervisors Miss Sarah Creighton and Mr Duncan Wilcox for giving me the opportunity to undertake this work, and for their guidance and support in completing the writing of this thesis. I am deeply indebted to Sarah Creighton who has not only supervised this doctoral thesis, but has become a mentor and friend.

I would also like to thank the Middlesex Clinic staff at University College Hospitals; Dr Lih-Meh Liao, Dr Gerry Conway and Sister Maligaye Bikoo for their ideas and general professional advice in tackling some of the more difficult issues involved in clinical research. I would also like to express my gratitude to Professor Christopher Woodhouse for his assistance in preparing this work for presentation at both national and international level.

In addition I would like to thank all of the participants who participated with such enthusiasm and demonstrated much interest in the research project.

Finally, I would like to thank my husband Ryan whose constant encouragement and little reminders ensured the completion of this written work.

PRESENTATIONS TO LEARNED SOCIETIES

Anorectal Malformations for the Gynaecologist.

Davies MC, Wilcox DT, Creighton SM

British Society of Paediatric & Adolescent Gynaecology, London, March 2004.

Adult Female Outcomes after Anorectal Malformation Repair.

Davies MC, Woodhouse CRJ, Wilcox DT, Creighton SM.

American Association of Pediatrics /European Society Pediatric Urology Joint Meeting, Uppsala, Sweden, June 2005.

Complex Vaginoplasty in Young Women.

Davies MC, Woodhouse CRJ, Creighton SM.

American Association of Pediatrics /European Society Pediatric Urology Joint Meeting, Uppsala, Sweden, June 2005.

Adult Female Outcomes after Anorectal Malformation Repair.

Davies MC, Woodhouse CRJ, Wilcox DT, Creighton SM.

British Association Urological Surgeons Annual Meeting, Glasgow, June 2005.

What are the Adult Outcomes after Anorectal Malformation Repair?

Davies MC, Woodhouse CRJ, Wilcox DT, Creighton SM.

American Urological Association Annual Meeting, Atlanta, USA, May 2006.

Genital anomalies and sexual function in males with Anorectal Malformation.

Davies MC, Woodhouse CRJ, Wilcox DT, Creighton SM.

British Association of Urological Surgeons Annual Meeting, Manchester, June 2006.

Gynaecological Outcomes after Anorectal Malformation (ARM) Repair in Childhood.

Davies MC, Wilcox DT, Creighton SM.

British Congress of Obstetrics & Gynaecology, London, July 2007- Winner of Best Poster Presentation Prize.

Chapter 1- Introduction

Introduction

Anorectal malformations (ARM's) and cloacal anomalies are rare and complex malformations of the lower gastrointestinal and genitourinary tract. They affect approximately 1 in 3500 -5000 live births (1). This represents approximately 180 new cases per year in England and Wales (2). They may occur in isolation or in association with abnormalities of the urogenital, cardiovascular, skeletal systems as with the VACTERL association, which is a presence of three or more anomalies in vertebrae, cardiac system, trachea, renal tract, oesophagus and limbs. The treatment of these patients has traditionally focused on achieving urinary and faecal continence, with preservation of renal function. In those patients where faecal continence is not possible diversion of the bowel may be required. It will often be possible to create a continent bladder though emptying may require intermittent catheterisation. A continent rectum is more difficult and many affected individuals remain clean only by a process of 'controlled constipation' or continue with a colostomy (3). Faecal continence remains a challenge and is achieved in about 60%.

The most severe form of cloaca is rare. It is often associated with other major congenital anomalies especially of the cardiovascular system. With all three channels coalescing (see figure 1), there is almost never an anal sphincter and seldom a urethral one. In intermediate forms the urethral sphincter is normal. As there are no endocrine association, the internal genitalia are often normal. The urinary anomalies are potentially lethal and the reconstructive surgery in childhood is technically very difficult. There is, therefore, little information on the long-term outcomes. The condition must be distinguished from cloacal exstrophy which is a severe variant of ectopia vesicae. With improved surgical technique and paediatric intensive care facilities these patients now live relatively normal lives, with a near normal life expectancy.

Background to Research Question

This research was prompted by surgeons treating patients with anorectal malformation as children and adolescents who were increasingly finding themselves unable to

answer specific questions with regards to long-term outcomes. Furthermore, the management of these patients is becoming increasingly holistic, and all aspects of the patients' outcomes are being considered. This research investigates all aspects of patient care and outcomes, including psychological, sexual function, fertility and paternity outcomes as well as the traditional bowel and urinary function.

Advances have been made in the pre-natal diagnosis of anorectal malformation (4), but at present the majority of cases are not diagnosed until after delivery. One of the main benefits of making an antenatal diagnosis is the possibility of diagnosing other major abnormalities should they exist, and allowing better management of the delivery and early surgical intervention. This should ideally take place in a tertiary referral centre with a fetal medicine department and paediatric surgery and neonatal intensive care facilities close to hand. The most common findings on prenatal ultrasound are bowel dilatation, ascites, polyhydramnios and hyperechoic intestine (5;6). However there are data to suggest that some of these radiological findings may be seen in normal fetuses (7), therefore complicating the issue of antenatal counselling of the parents. In one recent review of cloacal anomalies diagnosed prenatally it was found that the average fetal age at diagnosis was 27 weeks (8).

The reality of the situation is that the majority of patients seen in our unit are not diagnosed prenatally but are transferred after delivery. This is a difficult time for most families as the unexpected diagnosis of a major congenital abnormality in a newborn causes distress to the parents and family. Information provided at this difficult time may not be fully absorbed and may need to be repeated and offered in different media forms. However, if the information requested is lacking this can make a difficult situation impossible. The majority of data currently quoted when considering the long-term outcome of anorectal malformation is taken from studies based in the USA. Whilst this information is valid, in so much as the patients have undergone similar operative procedures there are nevertheless differences in the healthcare systems of the USA and the UK which may affect outcome results. This bias is most likely to be patient selection bias, that is those units publishing their results are likely to be larger, well funded research institutes, and therefore would attract not only the local population but also those most able to afford to be selective

in their treatment choice. This is in contrast to the UK where most people have little or no choice about the centre in which they receive their treatment.

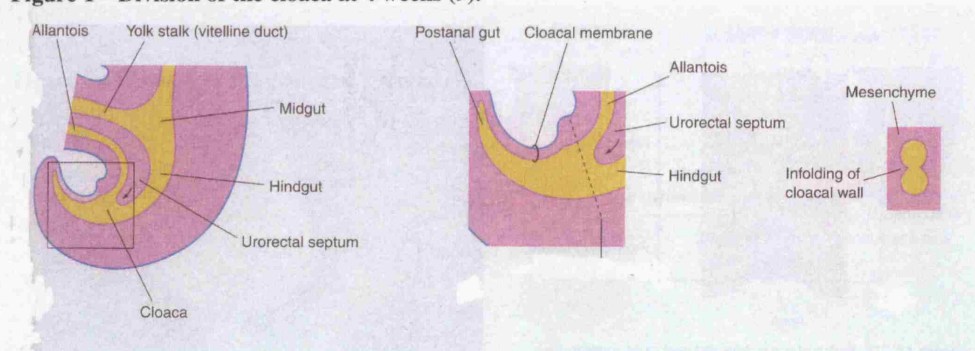
Development of a comprehensive database of patient outcomes for all of the children treated at Great Ormond Street Hospital (GOS) who are now adults would allow this gap to be filled. It would allow a range of outcome measures to be considered on the full spectrum of anorectal malformations. Much of the current published data focuses upon bowel and urinary incontinence, however sexual, psychological and fertility problems also exist.

Embryology

The normal human embryological development of the anorectum is a controversial subject. Rathke first describe the process of division of the cloaca as a result of fusion of the lateral folds in 1832. This fact was disputed by Tourneux in 1888 who stated that the urorectal septum descended in a coronal fashion. Most of the published work in the 20th century uses either or a combination of these two theories to account for the development of the perineal structures.

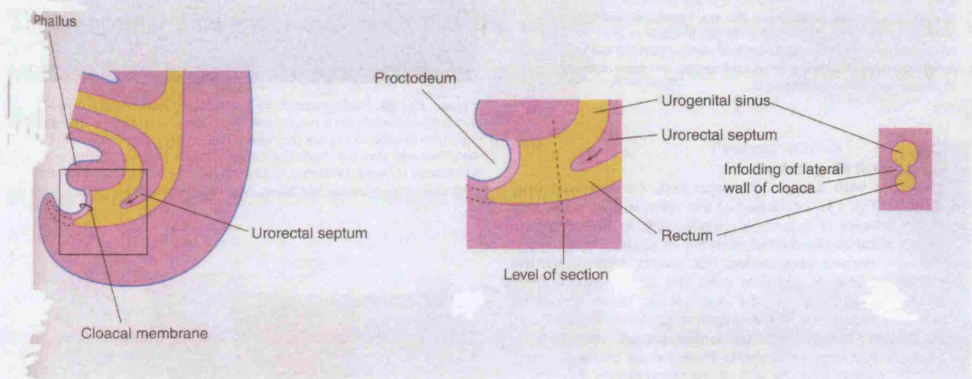
The distal portion of the primitive gut forms an expansion called the cloaca, this is in contact with the surface ectoderm at the cloacal membrane (see figure 1). Between weeks 4 to 6 of development the cloaca is partitioned into an anterior urogenital sinus and a posterior rectum.

Figure 1 – Division of the cloaca at 4 weeks (9).



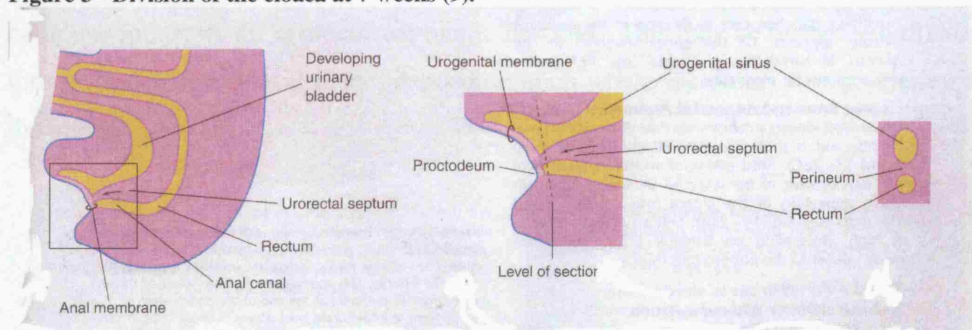
The division is due to growth of a coronal partition called the urorectal septum. As the septum grows toward the cloacal membrane it produces forklike projections that result in the lateral wall infolding (see figure 2).

Figure 2 - Division of the cloaca at 6 weeks (9).



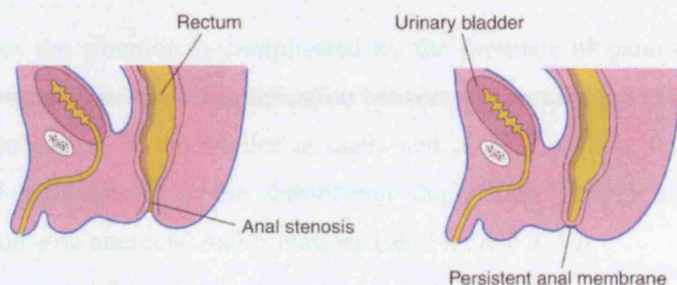
These lateral folds grow inwards and fuse forming the final partition that divides the cloaca in two. By week 7 the urorectal septum fuses with the cloacal membrane, dividing it into a dorsal anal membrane and a ventral urogenital membrane (see figure 3). The area of condensation between the urorectal septum and the cloacal membrane becomes the adult perineal body. Mesenchymal proliferation around the anal membrane forms a raised border – the anal pit. The anal membrane which has been separating the endoderm and ectoderm breaks down in week 8, and brings the digestive tract into communication with the amniotic cavity.

Figure 3 - Division of the cloaca at 7 weeks (9).



It is suggested therefore that anorectal malformations occur as a result of abnormal development of the urorectal septum causing incomplete separation of the cloaca. Low anorectal anomalies are those that lie distal to the puborectalis muscle. Examples include anal agenesis with or without a fistula (see figure 4). Essentially this group of ARM includes anal atresia, anal agenesis, ectopic anus and anal stenosis. The abnormal anal canal may open into the vagina in females and into the urethra in males. The majority of low anorectal anomalies are associated with an external fistula.

Figure 4 –Low ARM, anal stenosis and anal atresia (9).

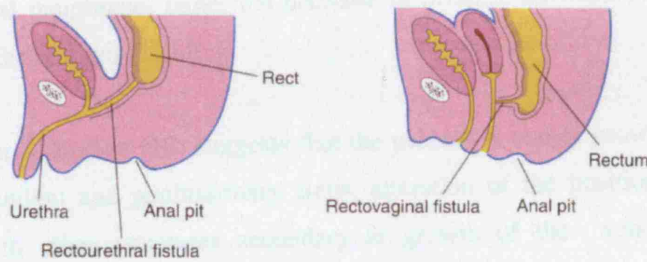


Anal stenosis is probably caused by some dorsal deviation of the urorectal septum resulting in a small anal canal. Anal atresia is probable the result of rupture of the anal membrane at the end of the eighth week of development.

In high ARM the rectum ends superior to the puborectalis muscle, there is usually an associated fistula to the bladder, vagina, vestibule of the vagina or urethra (see figure 5). The most severe defect is a result of a persistent cloaca is characterised by the complete failure of the urorectal septum to descend. This may be further sub-divided into high and low cloacal defect depending upon where the defect lies in relation to the pelvic floor.

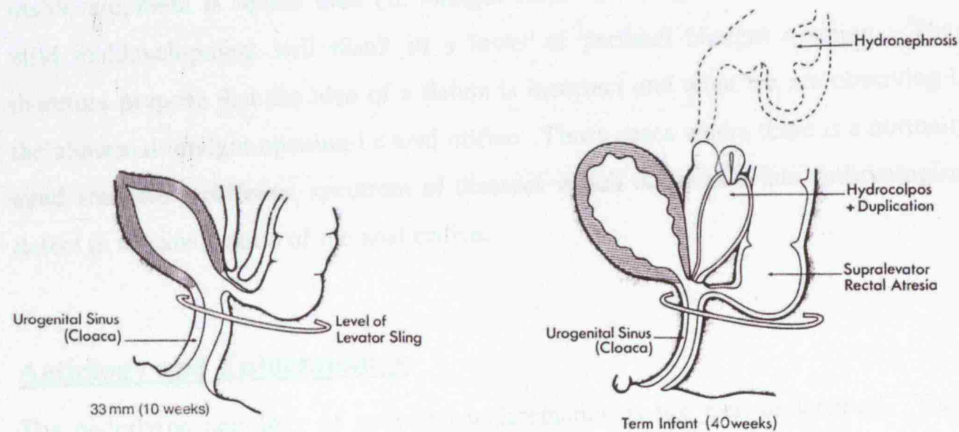
Figure 5 – High ARM anorectal malformation (9).

Two diagrams demonstrating high ARM in a male and female, with a fistula to the urethra (male) and vagina (female).



In females the situation is complicated by the presence of paramesonephric ducts. Any abnormal fistulous communication between the rectum and urogenital sinus may prevent migration of the Mullerian ducts and prevent midline fusion. This is the proposed mechanism of the subsequent duplication genital anomalies seen in association with anorectal malformations (see Figure 6) (10).

Figure 6 - Mullerian Duplication as a result of Cloacal Anomaly (11)



More recent research by Nievelstein et al state that the development of the human anorectum can be divided into two distinct periods; early and late (12). The early period correlates to the first seven weeks after fertilisation, during this time the initial structure of the hindgut and urogenital sinus is laid down. The later embryonic period corresponds to weeks 8-12 of gestation and is characterised by definitive organ

differentiation, the rupture of the cloacal membrane at day 49 is considered to be the point between these two periods. This study confirmed that during the caudal folding process parts of the yolk sac and allantois are incorporated into the embryo. This mesodermal tissue fuses to form the urorectal septum. This septum does not fuse with the cloacal membrane, rather the decrease in distance between the two is largely a result of the embryo infolding.

Penington & Hutson (10) suggests that the process is one of growth of mesenchyme of the hindgut and genitourinary sinus, alteration of the position of the cloaca in relation to other structures secondary to growth of the ventral infraumbilical abdominal wall, changes in curvature of the spine, and apoptosis of the dorsal wall of the cloaca, with shortening of the dorsal cloacal wall.

With these changes in our understanding of the embryological development of the normal human anorectum the underlying process of pathology that occurs in anorectal malformations cannot be adequately explained by the previously accepted theory of inadequate midline fusion or descent. Nievelstein proposes that ARM's occur as a result of inadequate development of the dorsal aspect of the cloaca and cloacal membrane, consequently the hindgut opening becomes anteriorly placed. If this maldevelopment is severe then the hindgut enters the urogenital sinus high up and mild maldevelopment will result in a lower or perineal hindgut opening. They therefore propose that the idea of a fistula is incorrect and what we are observing is the abnormal hindgut opening i.e anal orifice. Those cases where there is a normally sited anus are a different spectrum of diseases which occur as a late embryological defect in re-canalisation of the anal orifice.

Aetiology and Epidemiology

The underlying aetiology of anorectal malformation is not well understood. There appears to be no single aetiological cause to account for anorectal malformations, and this would seem to fit with the spectrum of anomalies encountered. In the largest epidemiological study looking at patients with anorectal malformation Cuschieri found that out of 1,846 patients with anal anomalies, 64% had other defects (13). In this large review 15% of cases had underlying chromosomal abnormalities, of which

Trisomies 21, 18 & 13, formed the largest group. A further 15% of babies were found to have VACTERL associations. Czeizel, suggests that babies with VACTERL were more likely to be male and of low fetal weight (14).

More recent research postulates the possibility of a genetic component with affected individuals having a 2 to 3 times higher rates of having another affected family member (15). Other suggested aetiologies include environmental factors such as fetal alcohol syndrome, infectious factors e.g. toxoplasmosis and cytomegalovirus. There is some evidence to suggest that folic acid taken during pregnancy may decrease the rate of anorectal malformation seen in the general population (16). However this data is the result of a population study and not from a randomised controlled trial, therefore the possibility exists that other factors may be protective in the folic acid taking population to decrease the rate of anorectal anomalies.

The remaining cases of ARM with associated anomalies fall into a miscellaneous group of Multiple Congenital Anomalies (MCA), which account for 60% (17). Martinez-Frias et al (18) have suggested a developmental field theory to account for the range of associated anomalies that may be present in these patients. This theory suggests that the entire embryo is subjected to an insult, and that the subsequent reaction is in an organised and integrated manner. Therefore isolated anomalies may be seen in those individuals where the insult occurred late in blastogenesis, and those individuals with multiple anomalies often in different systems are likely to have been exposed to an insult early in blastogenesis.

Classification

The most widely utilised system for the classification of ARM's is the Wingspread classification (19). This system distinguishes between high, intermediate and low ARM in males and females, and has a sub-group for cloacas and rare malformations. The level of the malformation is related to the level of the levator ani muscle, with high malformations lying above this level, intermediate at the level of the muscle and low malformations below this level. However, this classification system has been criticised as some of the types of malformation described are rare e.g. rectovaginal fistula. It is likely that the inclusion of this as a separate entity is may be a result of incorrect diagnosis rather than a change in the patterns of malformations that we are seeing. More widely used in recent years is the classification proposed by Pena (20) according to the type of fistula (see table 1). The benefit of this classification system is that not only does the type of fistula give information about the level of the anomaly but also to the extent of mobilisation required to treat, although with the introduction of the Posteriosaggital anorectoplasty (PSARP) procedure this has become less of an issue.

Table 1 – Pena Classification of Anorectal malformations

FEMALE	MALE
Perineal fistula	Perineal fistula
Vestibular fistula	Rectourethral bulbar fistula Rectourethral prostatic fistula Rectovesical (bladder neck) fistula
Imperforate anus without fistula	Imperforate anus without fistula
Rectal atresia and stenosis	Rectal atresia and stenosis
Cloaca	
Complex Malformations	Complex Malformations

Perhaps more important than the details of the classification system used is consistency of terminology. This is perhaps the single most important limiting factor when attempting to compare the outcomes of different units /surgeons. Therefore before considering any long-term follow research a consistent and robust means of

assessing patient outcomes should be considered. At present no single assessment of ARM exists therefore a combination of measures is required. It is important that any assessment tool that is used is one that is of international standing and has been validated. Of particular importance are the methods of assessing and reporting of urinary and faecal incontinence.

Existing Long-term Outcome Data

As stated previously, much of the existing data pertains to children or adolescents, and not to adults. Furthermore it is largely published from North American groups. The focus of much of this work is on faecal and urinary incontinence.

Urinary Function

In contrast to anorectal dysfunction, vesicoureteric dysfunction has the potential to cause permanent damage to other organs. Due to the long-term sequelae of upper urinary tract damage one of the main aspects of ARM treatment includes preservation of renal function and prevention of urinary tract infections (UTI's). Possible causes of urinary function may arise from:

- ARM fistula to the lower urinary tract
- Neuropathic bladder; primary or secondary to spinal anomalies
- Congenital abnormality of GU system
- Iatrogenic injury

Associated urinary tract abnormalities occur in approximately 40% of patients with a diagnosis of ARM (21-23). Commonly occurring problems include; reflux, renal dysgenesis and ectopic kidney (24). Furthermore the presence of two normally functioning kidneys is a positive predictive factor for future urinary continence (25).

Urinary tract problems may also arise secondary to iatrogenic injury. Reports have been made of urethral tears, urethral strictures, vas deferens injuries and ureteric injuries. The highest incidence of iatrogenic injury seems to be in males with a high ARM undergoing a pull-through procedure, where the reported rate in one series was 11% (26). The posterior sagittal approach seems to have improved matters, and rates of iatrogenic injury in a group of 572 patients operated upon in a single centre are

reported as being 3.3% (27). The two commonest injuries being urethral trauma, and inadvertent opening of the seminal vesicles.

Bowel Function

There is much written about the faecal continence outcomes after ARM repair. The general consensus seems to be that the most likely end result after PSARP is constipation (28). Most of the studies published looking at rectal function post-operatively use Kelly's clinical score to evaluate faecal continence, which does not have "constipation" as an outcome parameter. Pena proposes the underlying cause of constipation to be "...a hypomotility disorder of the rectosigmoid", this, he postulates, then allows the sigmoid to distend and lead to megasigmoid and overflow incontinence (29).

The bowel management of patients with ARM usually involves a combination of aperients, enemas and dietary manipulation, in order to achieve social continence so that patients are able with assistance, to be continent for the majority of the day. This usually requires some degree of colonic cleansing and reducing peristaltic action of the colon between enemas to avoid the passage of stool. In cases where there is failure to achieve continence the patient may be offered a permanent colostomy, or an antegrade continence enema (ACE) procedure. This latter procedure allows patients to self administer enemas via a small opening in the caecum which is connected to the anterior abdominal wall via the appendix. This is used to avoid chronic constipation and the overflow incontinence that ensues, and gives the patient a degree of autonomy in their management. There has been work on biofeedback as a possible solution with poor results in ARM patients. (30)

Unfortunately there is no uniformity amongst the medical literature as to the best way to assess and report patient outcomes. In an attempt to clarify the matter a summary table of results from a selection of papers found on a literature search has been included (Table 2). Papers selected were clear on the total number of patients treated and their outcomes. In order to compare the results produced from different centres three simple outcome parameters have been employed; good, fair and poor. Therefore in the case of Pena & Hong (31) where patient bowel function is discussed as being

continent or constipated, continence has been put into the “good” category and constipation into the “fair” category, as patients who are constipated may require enemas and experience overflow leakage etc. A more recent review of patients with cloaca by Pena et al has also been incorporated (32). Here the outcome of faecal incontinence has been classified as “poor”, occasional soiling as “fair”, and totally continent patients were placed in the “good” category. In papers by Hendren and Warne et al, spontaneous bowel movement was considered as a good outcome, regular enema administration was considered to be fair, and soiling or colostomy was considered a poor outcome. Even though patients with stomas are “continent” rectally, it cannot constitute a good outcome compared with spontaneous bowel movement.

The patients with high ARM tend to have the worst outcome with regards to faecal control. From the collective numbers in Table 2 only 28% (59 out of 213) of the high ARM group archived a good outcome, compared with 36% (122 out of 342) of cloaca repairs. The low ARM group fairs best with 83% achieving a good outcome and only 15% experiencing a poor outcome. In a study by Hassink et al, it is reported that none of their patients achieved “...normal continence for faeces” (33). The group also reports that 91% of the patients interviewed developed their maximal bowel control by the age of ten years. The effects of faecal incontinence on the patients in this series are reported mostly in an anecdotal manner but they do highlight the fact that almost 90% of the patients recall problems during school years with sporting activities. In a review of 1,192 patients in 2000 by Pena and Hong (10) it was reported that 75% of all their patients had voluntary bowel movements, with half of this group still soiling occasionally, therefore only 37.5% could be considered as being totally continent.

Table 2– Summary of Bowel Function results (34).

Literature review of bowel function outcomes, numbers given are total number of individuals included in each study.

ARM Type	Author	Good	Fair	Poor
Low	Iwai et al 1998 (35)	18	0	0
	Ackroyd & Nour 1994 (36)	46	1	1
	Pena & Hong 1998 (31)	47	18	0
Intermediate	Iwai et al 1984 (37)	10	3	0
	Ackroyd & Nour 1994 (38)	3	1	1
	Liem & Hau 2001 (39)	19	10	0
	Tsuji et al 2002 (40)	11	6	1
	Pena & Hong 1998 (31)	17	10	0
High	Iwai et al 1984 (37)	7	7	2
	Ackroyd & Nour 1994 (41)	11	6	10
	Liem & Hau 2001 (42)	9	8	2
	Tsuji et al 2002(40)	4	10	3
	Pena & Hong 1998 (31)	28	106	0
Cloaca	Hendren 1998 (43)	82	38	16
	Warne et al 2003 (44)	14	16	20
	Pena et al 2004 (32)	26	68	62

Gynaecological Outcomes.

Female Sexual Outcomes

There is no published information on the difficulties, if any, these females encounter during sexual intercourse or achieving sexual pleasure. Sexual difficulties, particularly related to anal dysfunction were found in 13% of those born with a low anomaly and 30% of those with a high or intermediate anomaly. The gender of the patients was not specified (45;46).

Genital Anomalies and Menstruation

Aside from the fistula, the associated female genital anomalies have been poorly documented but are known to be common. In a retrospective review, Hall et al found that the vagina had been assessed in only 72 of 162 girls and the internal genitalia in only 51. They found that 22 (of 72 - 32%) had a vaginal anomaly and 18 (of 51 – 35%) had a uterine anomaly (47). Subsequent studies have found the rate of female genital anomalies to be lower (48), this may be in part explained by the delayed diagnosis of female genital anomalies as many do not present until puberty.

In females with cloaca who survive into adult life the pattern of genital anomalies is similar. In ten of 41 girls a diagnosis of absent or vestigial uterus had been made on laparotomy in infancy, but six of them developed normal menstrual function at puberty: an observation that should make the paediatric surgeon very reticent in commenting on the internal genitalia (44). The authors of this study further recommend that all females with cloaca should undergo further gynaecological assessment early in puberty.

In those who have undergone vaginal surgery as part of their reconstruction, scarring is a common problem and can lead to stenosis and dyspareunia. The increasing recognition of the efficacy of vaginal dilation has made this the initial treatment of choice (49) as failure with this technique will not have had a detrimental effect on vaginal/perineal tissue. Nevertheless there are cases where it is almost impossible to provide an adequate vagina without transferring healthy tissue such as bowel or skin.

Hall et al report that eight of 17 patients for whom enough information was available had vaginal scarring and 'impaired vaginal function'. It was not known what effect this had on intercourse. It was felt that much of the problem lay in the unoestrogenised tissue that was operated upon in infancy. They recommended the deferment of vaginoplasty to the post pubertal period if possible (47). In those patients who have undergone repeated vaginal operations the failure rate of subsequent operations is unfortunately high. In a recent series stenosis occurred in 40% of patients who had undergone more than one vaginal reconstruction (50). If the vagina is too small, penetrative intercourse is impossible, but some patients prefer to use non-penetrative techniques rather than risk worsening their already tenuous anal control.

Pregnancy

Where intercourse is possible and the mullerian structures normal, pregnancy is possible. Many of those looking after ARM patients in pregnancy are concerned about the potential damage that could occur during the course of pregnancy and childbirth.

These include:

- Possible deterioration in renal function
- Worsening urinary incontinence
- Damage to urinary diversions during delivery
- Decreased faecal control as a result of pelvic floor injury

There are little other than case reports when considering pregnancy in patients with anorectal malformations (51). Iwai et al reported from their series of five females patients that only one had had a normal pregnancy by vaginal delivery and was married (52). One very comprehensive study of female obstetric outcome after cloacal repair is by Hendren, who reports that out of 24 adult patients, 17 are sexually active, and 6 have had babies although only one of these was via a vaginal delivery (53). Greenwell et al looked at 20 patients who had undergone lower urinary tract reconstructions for a variety of underlying conditions (54), they reported there was no significant deterioration in the renal function in the 29 pregnancies these women achieved. Furthermore, two-thirds of the patients underwent Caesarean section; the reasons postulated for this were reluctance to risk possible damage to the repair and

the patient's continence mechanism, spinal anomalies which precluded this group from spinal analgesia and the benefits of a planned procedure with regards to available staff.

The issue of potential affected offspring may be a concern in pregnant females with an ARM. However, familial anorectal malformation is rare and at present no single gene has been implicated. There are numerous reports of cases with varying patterns of inheritance (55-57). Nevertheless, potential parents will often ask if screening is possible. At present it seems that the pick-up rate for all of the gastro-intestinal atresias is low. When found, they usually are identified through the associated anomalies. In a large series from France of 118,265 fetal ultra sounds, the sensitivity for anorectal agenesis was 8.2% and none of the 27 isolated cases was identified. The mean stage of pregnancy at which anorectal agenesis was identified, was 27.5 weeks (58).

Male Outcomes

Sexual Function & Fertility

Impaired sexual function is seen in adult male patients who have undergone surgical procedures on the prostate and rectum but appears to be a rare consequence of the surgery entailed in repairing anorectal malformation (59). Whilst sexual function may be easily assessed by questioning the patient, fertility requires closer investigation of the patient with semen samples and imaging of the vas deferens. Therefore in most of the published literature we see paternity being used as a means of assessing fertility. There is little comprehensive data on this subject matter. A review by Pena identifies 20 male patients who have undergone puberty, all of whom are able to achieve an erection, 14 of these stated that they enjoyed masturbation and 6 had experienced sexual intercourse (31).

Male Genital Anomalies

Reasons for this group to be infertile include associated genital anomalies such as maldescent of the testes, associated sacral anomalies or iatrogenic injury during surgical repair. A study that looks more closely at this group by Holt et al found that in 20 patients investigated for infertility who had undergone an anorectal pull through

procedure that iatrogenic injury accounted for at least half of the cases (60). Other significant causes included recurrent epididymitis and associated anomalies.

Psychological Outcomes

In spite of improved surgical technique there may be an appreciable amount of psychological morbidity associated with anorectal malformation even after repair. This may result in significant emotional and social difficulty for both the patients and their families. The consequences of diagnosis of a serious medical condition at birth combined with protracted and repeated hospital admissions will undoubtedly have an effect on both the patients and their family. Increased levels of behavioural and psychological problems are known to exist in patients with chronic disease (61). It is therefore no surprise that patients with ARM also have increased rates of significant emotional problems.

In a report by Diseth & Emblem (62), 33 adolescent patients were looked at, and the rate of psychiatric diagnoses was found to be 58% compared with a rate of 35% in the group studied by Ludman et al (63). The differences between these two groups the author felt may have been due to the twice daily anal dilatations that these patients underwent as children. The intrusiveness of such a procedure may have had long term consequences for self esteem, body image and parental relationships, as it was usually the parent that had to administer the regime. It may be prudent to bear this in mind when considering bowel management regimes for young patients with ARM, where rectal enema administration may be required to achieve faecal continence.

Interestingly, it has been shown that the presence of faecal incontinence is not necessarily related to the degree or level of psychological maladjustment (63). This finding was not repeated in a later study by Bai et al (64) who reported that 66.7% of children with poor faecal continence had behavioural problems compared with only 8.6% in those considered to have good faecal control. They postulated that the effects of faecal incontinence would have a knock-on effect on the adult life of the patient including future occupation and relationships. More recent research has highlighted that there are certain factors which can be used to predict a poorer perception of health namely; increasing age, female gender and those with stomas (65).

Conclusions

The subsequent chapter will review the management of ARM looking at the types of surgical repairs these individuals may have undergone. It is essential that more detailed and specific information is available for surgeons and paediatricians involved in the care of these complex cases. Whilst data exists for many important aspects of these patients' outcomes including bowel and urinary outcomes, much of this originates from the USA, and may not be directly comparable to the UK population. In addition, little has been written specifically on sexual function and psychological outcomes in the adult population.

This study will focus on establishing the adult outcome data for bowel, urinary, sexual and psychological functioning. This study represents the largest cohort of British adults with ARM treated in a single centre, and aims to provide a comprehensive analysis of all aspects of their outcomes.

Chapter 2 - Treatment of Anorectal Malformations

Introduction

The majority of children born with an anorectal malformation will require urgent surgical intervention. The type of surgery usually depends upon the degree and level of the malformation. Many children undergo multiple surgical procedures to achieve social continence (urinary and faecal), this may carry on into adolescence and adult life. For those with a high ARM or cloaca the initial operative procedure is a colostomy to allow the passage of faeces whilst the definitive surgical procedure is planned. This allows the child a period of time to gain weight and for further investigation of other possible associated anomalies.

Prior to modern surgical and anaesthetic techniques of many of these children, particularly those with more severe anomalies would have died in the neonatal period, usually of overwhelming sepsis. The result of this is that there are more adolescents and young adults requiring management of these complex problems, and who may also require additional surgical interventions as they develop new problems either secondary to the ARM or of the associated anomalies.

Historical Considerations

The diagnosis of anorectal malformation or imperforate anus is not a new one, however there are very few early reports of the management of these patients, so most are assumed to have died without treatment. Paulus Aegineta (Paul of Aegina 625 – 690AD) is thought to have written the earliest account of a survivor of surgery for imperforate anus (66). His advice for treatment of this condition has been translated thus:

“In new-born children the anus is sometimes found imperforate, being blocked up by a membrane. If possible, then, the membrane is to be ruptured with the finger, but if not, we must cut it with the point of a scalpel, and accomplish the cure with wine” (67).

This can be thought of as the equivalent of a cut back operation, which is still performed today. One must bear in mind that these procedures were performed without the benefit of an anaesthetic or antiseptic agents.

Almost 1000 years later, in 1660, Scultet treated an infant with anal stenosis with dilatation. In 1676, Cooke used incision and dilatation and advised care of the sphincter muscles. In 1787, Bell suggested using a midline perineal incision to find the bowel. In 1783, Dubois acted on Littre's suggestion from 1710 by performing an inguinal colostomy for imperforate anus. Other surgeons followed suit, but almost all infants died, so colostomy remained unpopular and only a procedure of last resort. Formal perineal proctoplasty (mobilization of the bowel through a perineal incision and suturing it to the skin) was described by Amussat in 1835, and this technique gained rapid acceptance. Strictures occurred less commonly than with earlier procedures. In addition to Amussat, Dieffenbach also described anal transposition (1826); Chassaignac used a probe through a stoma to guide the perineal dissection (1856); and Leisrink (1872), McLeod (1880), and Hadra (1884) recommended opening the peritoneum if the bowel was not encountered from below.

An important development in the management of ARM was the introduction of imaging to delineate the abnormality. This was first advocated by Wangenstein and Rice in 1930 (68). Single-stage abdominoperineal procedures became widely used after reports by Rhoads, Pipes, Randall, Norris, Brophy, and Brayton (1948-1949). Stephens (1953) described this procedure and emphasized preservation of the puborectalis muscle. This surgery and its subsequent modifications were the standard approach until 1980.

Other contributing factors to the improved outcomes in ARM are the introduction of anaesthetic, antibiotics and the appreciation that children have improved outcomes if treated in specialist paediatric centres by paediatric surgeons.

Current Management

In some cases the management of anorectal malformation commences before the baby is born if the diagnosis is made antenatally. Preparation can then be made for the

child to be born in, or close to a specialist unit where paediatric surgeons and intensive care facilities are available. Initial management should include prompt screening for sacral and genitourinary anomalies. This can usually be achieved with an ultrasound of the urinary tract and lumbosacral radiography. In children where there is a significant spinal anomaly then MRI may be preferable and allows a more accurate assessment. Those found to have sacral dysgenesis should undergo urodynamic investigation in the first three months of life. Children who are seen to have dilated upper tracts may need additional investigation with a voiding cystourethrogram (69). The disability from renal impairment may in fact cause more problems in later life than the ARM, with some patients requiring renal function support or even a renal transplant.

Primary Surgery

For those children without a perineal fistula a colostomy is usually performed in the neonatal period. The child can continue to grow whilst awaiting definitive treatment, the colostomy also allows protection of the surgical repair post-operatively. There are however complications associated with performing colostomies on children. One study looking specifically at children born with anorectal malformation found that 30% of those treated with a loop stoma had complications and 70% of those with a divided stoma had complications (70). One recent large series looking at colostomy formation in neonates found the complication rate to be nearer to 80% (71), furthermore a mortality rate of 2.5% attributable to the stoma alone was also reported.

Treatment for patients with a high ARM or cloaca therefore consists of a minimum of three operative procedures. After colostomy formation, the main surgical repair takes place at approximately one month, or when the patient condition has stabilised sufficiently. This may be significantly delayed in patients with other serious congenital anomalies such as those requiring cardiac surgery.

PSARP

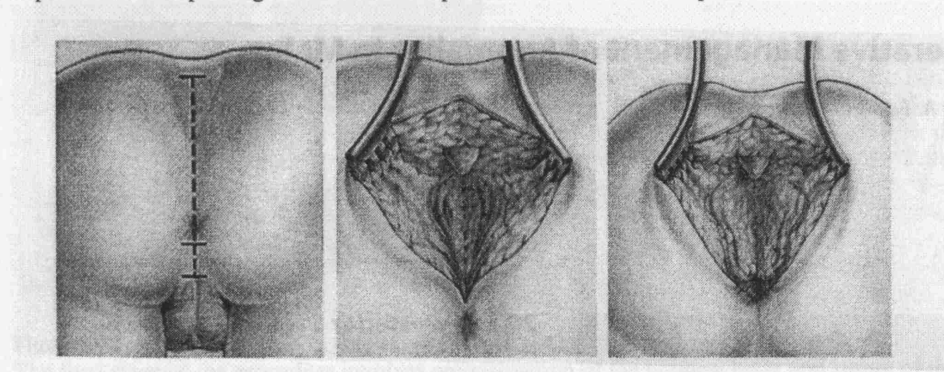
The surgical management of ARM has been revolutionised by the Posterior Sagittal Anorectoplasty (PSARP), which was first described by deVries and Pena in 1982 (72). This approach allows the operator direct access, and better visualisation of the

complex anatomy of the rectum and genitourinary tract via a posterior approach, which has improved surgical outcomes. Prior to this, children were treated with a variety of surgical procedures, depending on the level of the malformation. For low anorectal anomalies the procedure the most likely was an anal cutback. This involved making an incision along the perineum at the site of the rectum. The distal end of the rectum is mobilised down and sutured to the perineal skin. The limitations of this procedure are the poor visualisation of the anatomy, and the possible iatrogenic injury that may occur.

For the PSARP repair patients are positioned in the prone position. A midline incision is made and the parasagittal fibres are divided and the sphincter muscle complex is identified and divided, see Fig. 7. This process may be made easier with the use of an electrical stimulator to identify muscle contraction.

Figure 7 - Posterior Sagittal incision (73)

Separation of the parasagittal fibres and exposure of the muscle complex



The rectum is then divided from the urogenital structures and the fistula is identified and repaired, see fig. 8. The complexity of this stage of the operation depends enormously upon the level and type of malformation. When the rectum has been dissected sufficiently to allow it to be mobilised without tension to the perineum the size of the rectum should be evaluated. It may be necessary to taper the rectum before performing the rectoperineal anastomosis. The rectum should be placed within the sphincter muscle complex, and the sphincter repaired posteriorly, see figs. 9 & 10. All patients should be left with a urinary catheter and receive broad spectrum antibiotics post-operatively.

Figure 8 - Separation of the rectum from the urethra (73)

After dividing the muscle complex and levator muscle the rectum is completely separated from the urethra

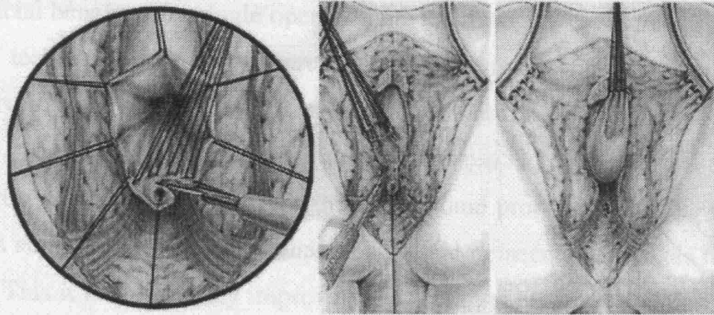


Figure 9 - The rectum is passed in front of the levator muscle (73)

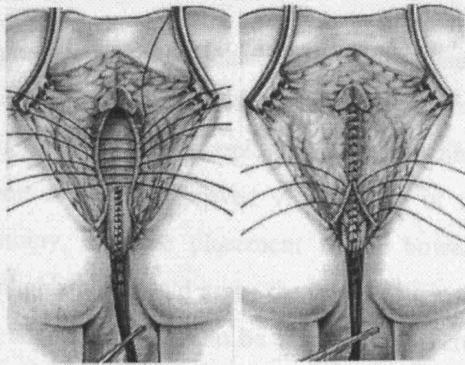
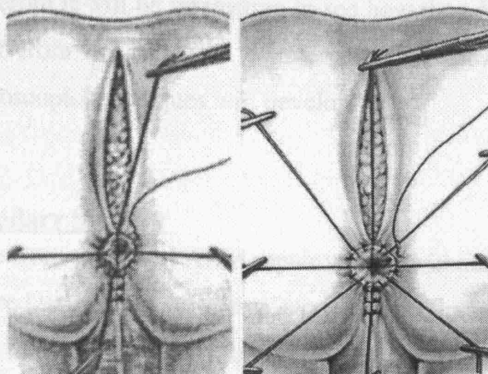


Figure 10 -Anoplasty (73)

The final stage of the procedure involves reconstruction of the perineal body and suture of the rectum onto perineal skin.



There are reports of some centres performing only a primary repair procedure, without a colostomy, therefore the child undergoes a single operative procedure (74). This has been found to be of particular use in developing countries, where the financial benefits of a single operative procedure are of even greater importance (75). This technique is not widespread probably due to the perceived increased risk involved. There are some that would argue that the risks were in fact decreased as there is only one anaesthetic, and there is less psychological burden upon the parents and rest of the family (76). Furthermore, some proponents of this procedure argue that a single early procedure improves normal defaecatory reflexes to be established (74). This it is argued may improve future continence. It may be that as more centres gain experience in this technique that it may become more popular and subject to a randomised control trial comparing its outcomes with the traditional three procedure approach.

Laparoscopic approach to the management of ARM has also been reported. Georgeson et al (77), describe the treatment of 11 patients with high ARM who underwent a laparoscopic assisted pull-through procedure. The advantages they report included improved visualisation of the pelvic floor musculature and fistula anatomy, accurate placement of the bowel, and a minimally invasive approach. Though some would argue that for the majority of patients treated there is no need for the abdominal cavity to be entered, and therefore the procedure is in fact maximally invasive, (comment by A Pena)(8). The potential advantages of laparoscopic procedures are reported to be improved vision of the pelvic organs allowing optimal placement of the anorectal pull-through and decreased perineal scarring (78). Therefore it will be interesting to see how the adult outcomes of these young patients differ from their PSARP counterparts, and how and when further introduction of laparoscopic techniques will develop.

Secondary Surgery

Aside from the initial ARM repair with or without a bowel diversion patients are often required to undergo repetitive surgical procedures and ongoing management into their adult lives. These may be related to the original ARM or any of the associated anomalies.

Bowel

Faecal continence is dependent upon three factors; sphincters, motility and sensation. Individuals born with an anorectal malformation may have problems with all of these mechanisms either as a result of their original anomaly or as a consequence of corrective treatment. Furthermore, impairment of sensation may have a negative effect on motility further compounding the problem.

For those patients in whom faecal continence is not achievable options for alleviating this problem include colostomy/ileostomy, antegrade continence enema (ACE) or a bowel management programme. Bowel management typically comprises of a combination of constipating agents such as loperamide and regular rectal enemas or washouts, thus allowing the individual to be socially continent (79). The aim of such treatment is to maintain faecal control without further surgical intervention, and many patients appreciate the opportunity to improve their situation without the need for further surgery. However when such protocols are instituted in the young with parents administering regular enemas or rectal washouts, this may lead to psychological problems. Furthermore, young patients requiring regular rectal washouts also experience problems with rectal mucosa prolapse and soreness, which may limit their ability to have a good result and compliance.

The commonest outcome following ARM repair is quoted as being constipation (80). This may manifest as pseudo-incontinence with faecal overflow rather than true incontinence (81). Therefore it is imperative that this is excluded as a treatable cause of incontinence before further surgical procedures are planned. If the patient is chronically constipated then they may require a bowel management regimen. Even if this is adhered to strictly there are cases where the terminal colon and rectum become grossly dilated, thus worsening the problem. In such cases the only option is bowel imbrication or resection (79).

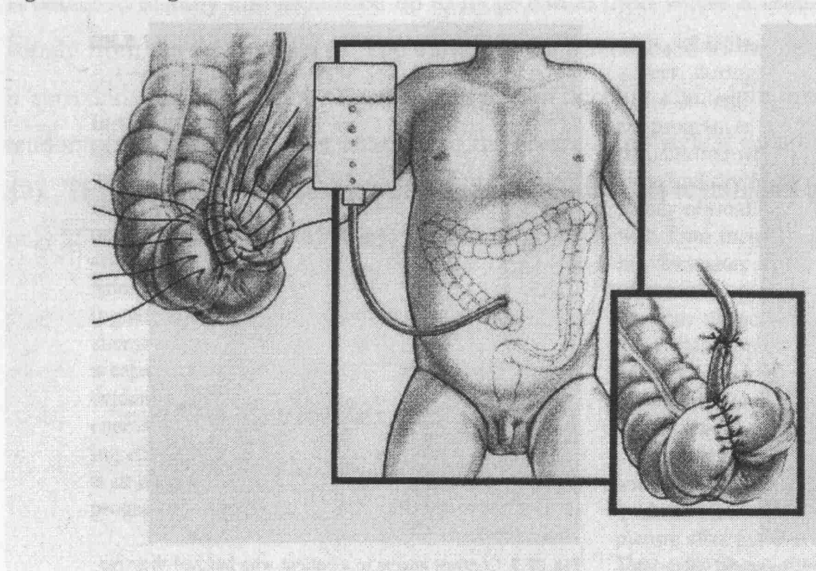
For a few patients the only means of achieving social continence is some form of stoma. This cannot be considered a good result and the patient should be duly counselled before hand. For many this is the final result after many surgical

procedures and it is not uncommon to find that patients are in fact rather glad to have alleviated themselves from the social stigma of being faecally incontinent.

Antegrade Continence Enema

For those patients who wish to avoid a colostomy or ileostomy, an ACE may be considered. This procedure was first described in 1990 by Malone et al (82), the authors highlighted the problem of rectal washouts in patients who had undergone multiple surgical procedures at the perineum and advocated antegrade washouts. The procedure involves creating a catheterisable channel to the caecum utilising the appendix to allow the administration of an antegrade enema, see figure 11. Where the appendix is not available, either after appendectomy or Mitrofanoff then two alternatives exist; these are the button caecostomy (83) and the use of a tubularised small bowel (84). There have been alternative procedures described but these are essentially modifications of the Malone procedure, usually entering a more distal segment of the colon (85). There are also reports of this procedure being performed laparoscopically with reasonable results (86;87).

Figure 11 – Antegrade Continence Enema Procedure (88).



The advantages of this procedure are that it gives the patient more control over their bowel washouts. This is of particular importance given that many of these patients

will as children and adolescents receive rectal enemas and washouts often administered by their parents (89).

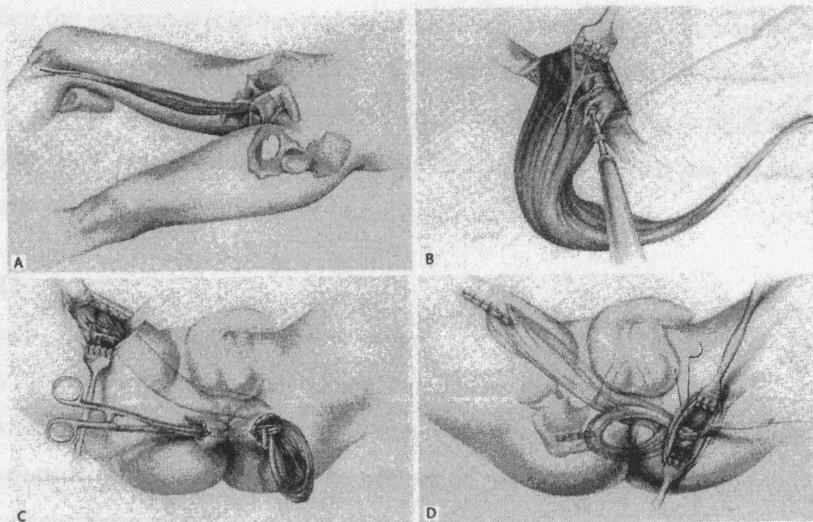
Recent outcomes data on this group of patients suggests that whilst significant problems with faecal control may remain even after a ACE, patient satisfaction with the procedure is high (90). Nevertheless, this is not a procedure without complication; prolapse, stenosis and leaking of the stoma have all been reported. One study looking specifically at medium to long-term outcomes of the ACE procedure found that in a group of 62 patients who had undergone an ACE procedure, 11(18%) were no longer using the stoma at a mean follow-up of 5.4 years (91). Reasons for this included lack of effectiveness, non-compliance and stoma complications. The authors also highlighted that problems were more likely in younger patients, and urged caution when considering this procedure in young children. Furthermore a co-existing diagnosis of colonic dysmotility is also thought to have a negative effect on the outcomes after an ACE procedure (92).

Graciloplasty

Conventional graciloplasty is the commonest surgical procedure for patients with persistent faecal incontinence and has been used since 1952 (93). The gracilis muscle is detached distally and mobilised up to its proximal third where it receives its nerve supply from the femoral nerve. The muscle is then transposed to the perineum where it surrounds the anorectum subcutaneously, then dorsally and again anteriorly. The tendon of the gracilis is then attached to the contralateral ischial tuberosity (see Fig. 12). The results of this technique in patients with ARM are reported as being good in only 25%, intermediate in 45% and poor in 30% (94).

Figure 12 - Gracilis Muscle Transplantation (95).

A Incision along medial aspect of the thigh. **B** Electrostimulation of the innervating branches of the femoral nerve. **C** Pull-through of the gracilis muscle to the perineum. **D** Fixation of the muscle to the contralateral ischial tuberosity



Dynamic Gracilioplasty

This technique utilises a transplanted gracilis muscle and an electrical stimulation device to improve faecal continence. The benefit of the electrical stimulation device is to promote fatigue resistant muscle fibres. The published result of this procedure quote success rates of up to 75% (96). However it is also recognised that individuals with high ARM do not fare as well with one study reporting improvement in only 18% and a complication rate of 57% (97).

Artificial Anal Sphincter

The artificial anal sphincter was developed in 1987 by Christiansen and Lorentzen (98) (AMS 800; American Medical Systems, Minneapolis, MN, USA). The use of this device is limited to those with severe faecal incontinence. There are data on its use in patients with other causes of faecal incontinence and its success rate is reported as being 66-80% (99;100). The only study looking at its use in ARM demonstrated an improvement in incontinence in all treated individuals. However the complication rate was high with 6 of the 11 (55%) treated individuals having at least one significant complication (101).

Permanent Bowel Stoma

In some cases of protracted faecal incontinence a permanent stoma may be the only solution. For many it is the end of the line after repeated surgical procedures, and the social continence that is achieved is often a positive event. However, for many including the surgeon it is often viewed as a failure. There are no accurate figures on exactly how many ARM patients end up with a colostomy or ileostomy, but it is likely that the number is small. Many of the issues faced by patients with a stoma are related to body-image and self-esteem however this is an area that has not been fully explored.

Urinary

Successful management of the lower urinary tract requires a low pressure bladder, healthy upper tracts and a functioning continence mechanism (102). In order to remain socially continent patients may be required to use clean intermittent self catheterisation (CISC). This may be performed per urethra or via a mitrofanoff, in some cases an incontinent stoma may be the only option.

Urostomy/ ileal conduit

An incontinent urinary diversion may be necessary for patients in whom continence cannot be established by any other means. This type of procedure pre-dates continent urinary diversion, and is usually seen in older ARM patients, although there may be circumstances which require this to be carried out. This allows the patient to be “dry” with a manageable stoma. Many of the early complications arising from this procedure are due to the ileal resection and subsequent anastomosis, and include anastomatic leak, enteric fistula, prolonged ileus and bowel obstruction. Specific conduit complications include necrosis, stricture, prolapse and retraction. Furthermore skin excoriation may result for ill-fitting stoma appliances. These patients require extensive pre-operative counselling as this is essentially an irreversible procedure as the native bladder is usually removed or shrinks down in size thus rendering it insufficient.

Neo-bladder formation & Bladder augmentation

As discussed previously urinary tract malformations often occur in association with ARM, particularly in the patients with cloaca. This may result in a small or

neuropathic bladder. One of the consequences of constant urinary incontinence for example due to an inadequate bladder neck is that the bladder does not stretch up enough to allow a normal capacity, so even if the incontinence issue is resolved there is often a problem with urinary frequency and incontinence. This may require the bladder to be enlarged or completely reconstructed. Bladder augmentation is usually performed using a patch of bowel to increase the maximum volume of the bladder. However the use of colon in the augmentation or reconstruction may worsen faecal incontinence by disrupting the colonic physiology (103). Furthermore, the use of ileal segments may also have an adverse effect on the faecal continence by decreasing the consistency of the stools.

In addition to the continence issues there are also long-term consequences where bowel is used in bladder augmentation. These problems are often predictable and treatable and include; metabolic acidosis, calculus formation, and tumours. The semi-permeable nature of bowel allows ammonia absorption leading to an increased acid load, the consequent renal potassium wasting may cause hypokalaemia. This seems to be more of a problem where stomach and jejunum are utilised thus making these less favourable bowel segments for bladder augmentation (104). Stone formation is for some patients a recurring problem, and arises due to chronic bacteriuria, urinary stasis, mucous production, and associated metabolic abnormalities. The commonest stones are composed of struvite and are thought to be secondary to urea splitting organisms (105). The best management for stone formation is prevention. Patients should be encouraged to seek early treatment for infections, washout the bladder daily with sterile water to decrease the mucous inside the bladder and regular drainage of the bladder (106).

This procedure may be carried out in conjunction with other reconstructive operations such as a Mitrofanoff. The benefits are that the patient is left with a bladder that is mostly native and may be able to achieve near normal bladder function. The adverse effects that one sees in neo-bladder formation with small bowel may be seen to a lesser degree.

Mitrofanoff

The development of a catheterisable conduit is comparable in its impact on the management of incontinence comparable with the ACE. It has allowed many young adults and adolescents the independence to pursue a normal life without the inconvenience of trying to perform regular urethral catheterisation or requiring an ileal conduit or urostomy. The Mitrofanoff relies on forming a conduit from the anterior abdominal wall to the bladder, usually from the vermiform appendix. The conduit may be to the native bladder or neobladder as described above. A valve mechanism prevents refluxing of urine, and allows catheterisation to occur. Patients are required to undertake regular self-catheterisation of the bladder but are able to do so in a more convenient manner. The complications are similar to those encountered with any form of stoma, namely stenosis, prolapse, and retraction.

Genital

Genital abnormalities are frequently seen in patients with anorectal malformations, especially those with “high” lesions. The detected rate from a series of 128 patients by Metts *et al* is reported as 26% in males and only 5% in females (107). Anomalies most frequently detected in males were undescended testis and hypospadias, in females the commonest genital anomaly was duplicate vagina. The significant difference between the two groups they suggest may be accounted for by lower detection rates in females due to inadequate assessment. Abnormalities are normally detected during the post-delivery examination of the baby. In females diagnosis may not occur until puberty, when the patient presents with amenorrhea and/or haematocolpos.

Vaginal Construction

Many of the vaginal anomalies seen in conjunction with ARM are minor and can be dealt with minimal surgical intervention. In those cases of cloaca the situation is rather more complicated as the vagina, urethra and rectum form a single channel. Therefore surgery to the vagina is performed at an early age, usually when the major repair is being carried out.

It may seem curious to consider sexual outcomes when ARM patients are first diagnosed, but it has been postulated that normal sexual development is more

probable if it is expected and prepared for in childhood (108). As children born with these complex malformations are undergoing successful reconstructions and developing into adulthood pregnancy is an increasingly common event. Unfortunately there is little in the published literature on the consequences of pregnancy on the reconstruction and urinary tract and on the mode of delivery.

Conclusions

The management of ARM can be complicated and involves many surgical procedures. In addition to treatment of the physical aspect of these conditions it is important that psychological and emotional issues are addressed, particularly in those individuals who require repeated surgical procedures.

Chapter 3 – Methods & Techniques

Introduction

In order to assess a wide range of outcome measures a questionnaire based study was deemed appropriate. This allows a large volume of data to be collected in one attempt. Given the wide range of problems these individuals encounter, a number of questionnaires were employed, as a single disease specific outcome measure of ARM does not exist at present. Areas it was felt necessary to assess included urinary function, bowel function, sexual function and satisfaction, body esteem, and quality of life. Also included in the questionnaire packs were consent forms which participants were required to complete and sign to demonstrate their consent to participate in the study. Sociodemographic data was also collected, which included details of employment, level of education attained and ethnicity to see if there were factors other than the disease process which may influence outcomes. Treatment in hospitals other than the study hospital was also enquired about.

Information regarding the participants' initial assessment and diagnosis was collected from paediatric medical records. A comprehensive past surgical history was also sought as well as information about co-existing anomalies. In conjunction with the questionnaires patients were also requested to attend for an out-patients appointment to go over the questionnaires and complete any missing data. The appointment provided an opportunity for the participants to ask questions about the treatment they had received as children, as many were unclear as to the exact nature of their condition and operations they had undergone as babies and small children. Finally, consenting patients were examined, with height and weight being measured. The number and location of any scars and stomas were also documented. An examination of the external genitalia and rectum was also performed if consent was given.

All patients had undergone some of or all of their treatment at Great Ormond Street Hospital. They were identified from the hospital database. Diagnoses that were searched for included anorectal malformation, cloaca and imperforate anus. In

addition the theatre logbooks were searched for all babies and children undergoing procedures that may be associated with a diagnosis of ARM including; colostomy, anal cutback, pullthrough, anoplasty and PSARP procedures. Medical records of these individuals were sought and the diagnosis checked.

All participants were 18 years of age or over. Ethical approval was sought from Great Ormond Street and University College Hospital and granted. Exclusion criteria were limited to patients known to have concomitant learning disabilities as it was felt that there may be some difficulties in their ability to self-complete the questionnaires.

One of the difficulties faced when trying to assess patient outcomes are the potential for bias. This bias could be considered to be twofold. Firstly patient selection bias may occur when recruiting patients, particularly if these patients are recruited from within the hospital either as in-patients or out-patients. The very fact that they are in receipt of treatment or follow-up means that they may not be entirely representative of the patient group as a whole, and therefore exert some form of skew on the results. In order to overcome this all patients that were registered in the Great Ormond Street Hospital database with an anorectal malformation were contacted via postal invite to take part in the study. Patients were not recruited from the hospital population or support groups, to reduce the possibility of patient selection.

Secondly, there may be a non-response bias, those patients choosing not to respond to the postal invite may be in some respect different to those who do respond to the initial invitation to take part. It has been suggested that this group may differ from those patients who do respond may have higher rates of the illiterate, foreign or homeless patients (109). It has also been postulated that those patients motivated to respond to postal invitation questionnaire surveys are those who are dissatisfied with the treatment they have received. However it would seem that the converse is more likely to be the case. That is the patients who experienced the fewest problems or those who are most satisfied are the most likely to return a completed questionnaire, either because they feel indebted to the provider of the care or because it is easier to express praise or thanks than criticism (110).

Tracing & Recruitment

It is known that postal invitation to take part in medical studies has its limitations, and uptake can be poor. As many of these patients have been discharged from GOS for many years it was necessary to find an up-to-date address. The chances of this have been improved drastically by the introduction of the National Health Service (NHS) strategic tracing service. This is a nationwide database that allows registered users to trace patients. All persons registered with a general practitioner (GP) are placed on this database and their home addresses and GP details are available. However the minimum data set required to perform a search are name, surname and date of birth, if any of these are missing then a search is not possible. This system allowed many of the older patients who had moved since their last appointment at GOS to be traced and invited to take part in the study. Licence to access the NHS tracing system is via the hospitals Caldicott guardian.

All patients were sent an initial invite to take part in the study and a stamped address envelope for its return (see appendix 1). Patients were also asked if they would like further information prior to deciding whether or not to participate, in such cases a patient information leaflet was sent (see appendix 2). If no response was received then the address details were checked again via the NHS tracing system, and a repeat invitation sent.

Questionnaires

Detailed questionnaires were produced and are included in appendices 3 & 4. These covered:

- Bowel Function
- Urinary Function
- Sexual Function & Fertility
- Quality of Life
- Body Esteem

Bowel Function

Perhaps one of the more important aspects of functional outcome requiring assessment is bowel control. It is for many patients the single most important aspect of their condition that they would wish to improve. The majority of patients do not achieve *normal* faecal control, and what control they do have is normally achieved in adolescence (111). The implications of a childhood spent with faecal incontinence are immeasurable. Much of the literature written on ARM utilises the Kelly's scoring system (112). This is a tool specifically designed for use in children and does not include constipation as an outcome measure.

As patients may have problems with constipation and/or incontinence it was imperative that both of these were measured. As there was no specific tool to assess these symptoms together it was necessary to find surveys that would complement each other.

In addition the possibility of utilising anal manometry as part of bowel function assessment was considered. There are reports to suggest that there is a positive correlation between continence and the resting anal pressure profile (113). However there are also data to suggest that clinical findings and manometry data are contradictory (114;115).

Faecal incontinence may be for solid or liquid stool or for flatus. An assessment of the frequency and type of stool must be part of any assessment. Also included should be details with regards to the ability to defer defecation and the requirement for a pad or anal plug, as many patients manage their incontinence with coping behaviours such as being close to toilets and taking constipating medications. Pescatori et al devised a score that took into account both the degree and frequency of incontinence (116). However this system does not take into account the quantity of stool lost. A more comprehensive scoring system was developed by American Medical Systems (117), however this is a fairly complex tool to utilise, and as it was to be part of a collection of other scoring systems it was felt that a more simple system was required. Jorge & Wexner developed a system that took into account not only the consistency and frequency of incontinence but also the affect on lifestyle (118) and it is an adaptation

of this score that was incorporated into our patients questionnaires (119). The benefits of using this scoring system over others are that it has been validated on a British population, although its limitations are that this process was only undertaken in female patients. Nevertheless the questions do not appear to be gender biased in any way, though obviously further validation on a mixed population would have been preferable.

The other important aspect of bowel function assessment was constipation, as the majority of patients who have undergone repair of ARM are constipated (120;121). Constipation is a symptom that is difficult to assess as the pattern of normal bowel habit can vary from once every 3 days to 2-3 times per day. Infrequency may be due to many factors including diet, exercise and medication. The Constipation Scoring System was utilised as the means of assessing the degree of constipation (122).

Lower Urinary Tract Symptoms Questionnaire

Urinary tract anomalies are frequently associated with ARM, the incidence varies according to the location of the anomaly, and may be as high as 30 – 50 % in high ARM (123-125). The most common problem encountered is reflux (126), which may cause significant morbidity and even mortality. Therefore an assessment of urinary function was of vital importance in our assessment of the patients.

There are many tools available for the assessment of urinary function and symptoms. The aim of these is to attempt to distinguish between the various urinary symptoms and their effect on the individual. They may also try to discern between the varying types of incontinence and the degree of incontinence. Features that we felt important were that the questionnaire had been developed in English and had been validated in the UK.

The female urinary function questionnaire used in this study was the Bristol Female Lower Urinary Tract Symptoms Questionnaire (BFLUTS) (127). BFLUTS allows a qualitative and quantitative assessment of symptoms of the female lower urinary tract. The questionnaire consists of 34 questions; 9 on incontinence, 12 on other urinary symptoms, 4 regarding sexual function and 9 on quality of life. The BFLUTS is a

well-known questionnaire which has been widely used in other studies and validated on a UK population. This may be an important factor when reporting symptoms of an embarrassing nature, where a national difference may exist. Response scales for symptoms range from “none” to “always” with “no problem” to “a serious problem” for bothersomeness. It has been shown to have good reliability and psychometric validity. Furthermore, the questionnaire is simple to understand and takes approximately 20 minutes to complete, which should optimise the return rate (128). This questionnaire has been used before in our unit and therefore has the advantage of allowing us to compare results between different patients groups.

The male urinary function questionnaire developed by the same group is the International Continence Society (ICS) male questionnaire (129). This was specifically designed for use in men with outflow obstruction symptoms from benign prostatic hypertrophy. However as it was similar in design to the BFLUTS questionnaire and had been validated on a UK population it seemed the best option. Additional questions were also included on the any urinary diversions the patients may have and also whether they perform ICSC.

Quality of life

The concept that quality of life could be measured is a relatively recent one, and has only really been considered since the change in definition of health by the World Health Organisation (WHO). After World War II, the WHO defined health as, “a state of complete physical, mental and social well-being and not merely the absence of disease and infirmity” (130). Quality of life research fills an important role in clinical medicine as it allows a patient perspective to be measured as an outcome. For many patients with congenital conditions much of the published data focuses upon outcome measures as reported by physicians or parents, as children are often deemed not able to report accurately health outcome measures. Therefore an inclusion of this in our research was deemed imperative.

When considering assessing quality of life there are two main means of assessment. One is a generic measure of life quality with regards to physical function, symptoms, psychological and social well-being, cognitive functioning and personal constructs

(131). Examples of these include the Short Form Health Survey (SF-36) and the World Health Organisation Quality of Life (WHO-QOL) score. The others are disease specific assessments, which are specific to a single health problem or body site. The benefits of these are obvious however they make comparison between affected individuals and control groups difficult. The specificity of these measures allows one to monitor clinical effectiveness of interventions and to take into account an individual's perspective. As no specific quality of life scores exist for patients with ARM it was decided that a generic quality of life assessment should be employed.

The SF-36 is a general health questionnaire developed by the Rand Corporation's Health Insurance Experiment in the United States (132), and has been validated for use in the United Kingdom (133). It contains eight dimensions, plus information about any change in health that may have occurred over the last year. The items in the SF-36 are derived from the original 108 measures that were in the original RAND health insurance experiment. This shortened version is the most widely used generic measure of quality of life in the US and UK.

The SF-36 contains 36 items which measure eight dimensions; physical functioning, social functioning, role limitations due to physical problems, role limitation due to emotional problems, mental health, energy/vitality, pain and general health perception. Item scores for each item are coded and transformed on a scale to give an overall score from 0 (worst possible) to 100 (best possible).

Sexual Function

The importance of sexuality as a part of well being is well recognised. The WHO has identified three basic elements of sexual health (134);

- *The capacity to enjoy and control sexual and reproductive behaviour in accordance with social and personal ethics.*
- *Freedom from fear, shame, guilt, misconceptions and other psychological factors that inhibit the sexual response and impair sexual relationships.*
- *Freedom from organic disorders, disease and deficiencies that may interfere with either sexual or reproductive function or both.*

These are fundamental aspects of a person's well-being, therefore in any long-term outcomes study of disease it is logical that there is some form of assessment of sexual and reproductive function. These issues are often overlooked by paediatric services as the focus of treatment and outcomes is usually urinary and faecal continence. Nevertheless, as the child becomes an adolescent the issue of sexual function and reproductive potential becomes more important, especially as the issues of continence have usually been dealt with.

Assessment of sexual function has many facets and it is may be difficult to include all of these in a single assessment. There are many scores for assessing sexual function in adult populations. As many of the available scores require the person to be engaged in heterosexual relationship to complete, we wished to find a scoring system that could be used on people of all sexual preferences and for those who had not yet had penetrative sexual intercourse. The Multidimensional Sexuality Questionnaire (MSQ) (135) is a generic scoring system that can be used on males and females. Responses are recorded using a five point scale ranging from "not at all characteristic of me" to "very characteristic of me". Furthermore, it allows people who have not yet been sexually active to complete it based upon an imagined relationship. It has 12 subscales looking at:

- Sexual esteem
- Sexual preoccupation
- Internal sexual control
- Sexual consciousness
- Sexual motivation
- Sexual anxiety
- Sexual assertiveness
- Sexual depression
- External sexual control
- Sexual monitoring
- Fear of sex
- Sexual satisfaction

This study utilised an abbreviated version of this questionnaire which utilised three positive and three negative subscales; Sexual assertiveness, sexual esteem and sexual satisfaction were the positive scales used. Sexual depression, fear of sex and sexual anxiety were the three negative subscales employed. The aim of utilising an abbreviated version was to reduce the number of questions that participants were required to respond to.

In order to have complete data on our subject's questions were included on the age of sexual debut, pregnancies, if any and their results e.g. miscarriage, live-born and mode of delivery females, and paternity was assessed in males.

In addition to these it was felt appropriate to assess the physical functioning of the genital tracts, in females this was assessed by questions about the onset of menses, any operations required to facilitated menstruation and childbirth. In males this was assessed using the brief Male Sexual Function Inventory for Urology (136) . This assess five subscales of males sexual function; sexual drive, erection, ejaculation, problem assessment and satisfaction. Whilst some of these are already assessed in the MSQ, the more specific aspects of genital function were lacking. In the development of the questionnaire it was noted by O'Leary *et al* that the inclusion of ejaculatory function was perhaps the single most important inclusion when compared with other scales assessing sexual function, which were often more psychological in nature. Ejaculation and the subjects satisfaction with it is something they felt was an issue overlooked by many physicians and healthcare professionals, yet seemed to be an important issue to patients.

Body Esteem

Body esteem refers to the overall positive or negative evaluation of the body by and individual (137). Poor body esteem is felt to be prevalent in people with disabilities and chronic disease states. Furthermore this may be compounded by the fact that society as a whole perceives the bodies of the disabled in a negative way (138).

As many of the scoring systems were gender specific it was deemed necessary to find a tool that could be used in both males and females, to allow comparison to be made

between both groups. The only tool that seemed suitable for this was the Body Esteem Scale (139;140). This scale was developed specifically for use on young adult populations, and allows a means of not only comparison within our sample population but also with published norms. It looks at a range of body parts and aspects of bodily functioning and asks how confident the person is about each area or feature.

Discussion

There are many drawbacks to survey via postal questionnaires, namely the lack of individuality of the questionnaires and the large number of questions asked which may not be relevant to the individual. However, they do allow a standard amount of information to be elicited and guarantee that some seemingly irrelevant information is not left out. There are also those that advocate the use of questionnaires studies in cases where the information required is of a more personal nature (141), as patients may be more willing to divulge symptoms of an embarrassing nature in a self-completed questionnaire rather than to a doctor in a face-to-face consultation (142).

However, if there are a significant percentage of non-responders i.e. people sent the questionnaire but not returned, this may introduce its own bias. It may be that only those patients with problems see the questions as relevant and therefore complete and return it. Nevertheless, one cannot assume that only those with problems are likely to participate in outcome surveys, and one must presume that patients with problems are as likely to not return completed questionnaires. Therefore to minimise this it is important to maximise response rates, there are recommended strategies that have been shown to achieve maximal response rates and these were incorporated into our study design (143).

Simple measures that can increase response rates include; using shorter questionnaires, sending the questionnaire in a brown as opposed to a white envelope, enclosing a stamped address envelope for its return. Other measures that improved return rates included making contact prior to sending out the questionnaires and supplying an explanation for the research. Many of these techniques were used in this study.

Bowel Function

Other means of assessing bowel function include anal manometry, this would provide an objective measure of anal function. However, specialist equipment is required and it is a fairly invasive procedure. In the majority of previously published data manometric data has been interpreted in combination with clinical findings (37;113). Furthermore the possibility of the clinical findings and manometry data not correlating has been reported (114). Imaging to study anorectal function has also been employed in the assessment of long-term outcomes. Defecography has been utilised in some centres (144), again this was not deemed appropriate in our study as it is a fairly invasive procedure, and may deter participation in the study.

Quality of life

The mode of questionnaire administration and outcome score has been investigated. It has been demonstrated that scores are lower from postal responses when compared with interview administered questionnaires (145). The postulated reason behind this was patients tend to under-report symptoms particularly with regards to mental health when directly questioned by an interviewer. What is unclear is whether the results from self-reporting are more accurate than those from interviewing. Nevertheless, postal questionnaires are a cheap and convenient means of health research and audit, but may under-represent the effectiveness of health interventions.

Sexual Function

Other aspects that should also be assessed as part of sexual health are reproductive outcomes. These are of considerable importance, as there are a significant percentage of patients with anorectal malformations who have associated anomalies of the genitourinary system. Again whilst this may not seem of paramount importance at the time of diagnosis, it may become an issue in adolescence when sexuality, sexual function and fertility are deemed important (146;147). Furthermore, with improved patient information facilities and easier access to medical literature via the worldwide web, it is an increasingly more likely that parents will question physicians treating their children with regards to the potential long-term outcomes.

The advantages of assessing male fertility via semen sample was considered though it was felt that there were potential ethical issues with the disclosure of this information, for example in cases of azoospermia where the male subject has apparently fathered offspring. This could cause unnecessary disruption to the family unit, and patients would have to be counselled and consented very carefully prior to participation. Non-paternity rates in the general population are quoted as being in the region of 10% (148) and there is no reason to suspect that there would be any difference in this sample.

Body Esteem

Many of the studies looking at body esteem focus on patients with eating disorders and those who have had a spinal cord injury. Neither of these groups are comparable with our patients, and indeed there will be a range of heterogeneity in our sample as there will be a range of outcomes from those with minimal problems and scarring to those with stomas and chronic continence problems. One of the limiting features of this questionnaire is the fact that the published normal values are from American university students, which may not be the best population to compare our participants' outcomes to. It would have been more satisfactory if normal values for a British population existed, or if the questionnaire had been previously used in a study with individuals with similar problems as our individuals, such as Hirschsprungs disease.

Conclusions

Whilst no single tool exists for assessing adult outcomes after anorectal malformation repair, a combination of validated questionnaires can be used to cover a range of functional outcomes. It would be useful if future outcomes research in this patient group in the UK could try and utilise some of the same tools to allow a direct comparison in their outcomes status. This would be of particular benefit in groups that have undergone different surgical procedures.

Chapter 4 - General Results

Introduction

In total there were 284 patient details stored on the database as having a diagnosis of imperforate anus, anorectal malformation (ARM) or cloaca, and being over the age of 16 years. All of these patients were contacted and invited to take part in the study. 33 of the 284 were not able to be traced, the reasons for this were either the search produced too many results, usually a result of having a popular name, or the patient was not registered with a General Practitioner. In such cases no invite was sent, furthermore as ethics approval granted at the time of project registration was for patients over the age of 18 years, 15 subjects were too young to participate in the project. Other patients on the database were also unable to participate as they had passed away, moved overseas, or were found to subsequently have a diagnosis of bladder exstrophy (see table 3).

Table 3– Summary of Patients unable to participate

Reason	Number
Unable to trace	33 (56%)
Too young	15 (25%)
Died	5 (8%)
Moved Overseas	2 (3%)
Bladder Exstrophy	4 (7%)
TOTAL	59

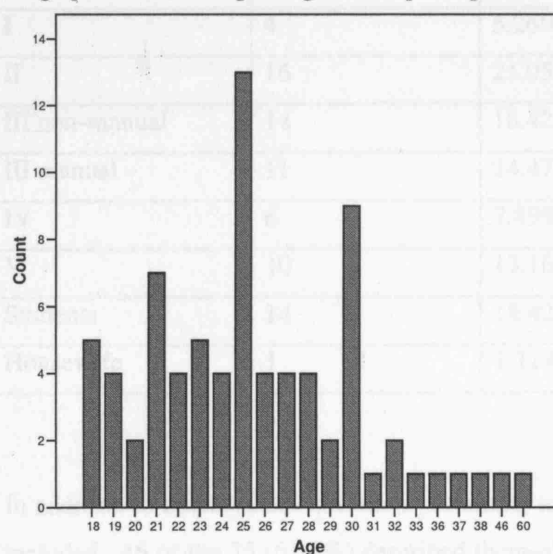
Therefore in total 225 postal invitations were sent to eligible participants, patients were requested to respond and a stamped addressed envelope was enclosed for their convenience. If no response was received after 4 weeks then a further letter was sent. In total 114 responses were received (50%), due to the nature of the NHS tracing system it is likely that some of the non-responders had moved to different addresses and had not yet registered with a new General Practitioner. This fact is made more likely due to the fact that the majority of the participants are young and therefore more likely to be changing address.

Of these 114 responses 95 agreed to take part in the study and were sent questionnaires to complete and 76 of these were returned completed. Those that did not complete and return their questionnaires were contacted and offered either assistance to complete the questionnaires or another copy. 15 patients stated that they did not wish to take part and 5 requested more information about the study and then declined to participate. Therefore out of the people that we were able to establish contact with 76/114 (66.67%) completed the questionnaires. One female patient was later excluded as she was found to have a diagnosis of exstrophy when her medical records were retrieved.

The age range of the participants was 18-60 years with a median of 25 years (see figure 4). The 60 year old was not on the GOS database but had been treated there for an anorectal malformation, he was recruited into the study via his daughter who also had an anorectal malformation. There were 44 females (59%) and 31 males (41%), the difference between the rate of male and female participants is not accounted for by any difference in the original database where the ratios were reversed and 44% were female and 56% male. The reason for this increased rate of participation by females represents a bias in the data however no simple explanation could be found to account for it.

Figure 13 – Age Distribution of Participants

Bar graph demonstrating the ages of the participants.



Demographics

Basic demographic data were collected on all participants. The vast majority (91%) described themselves as white British, the others were either British Asian, Indian, mixed race or white other. This is in concordance with current demographic data in the United Kingdom (149). When considering occupation the data was classified according to social grade as defined by the Office of Population Censuses and Surveys, however this classification does not have a division for students and parental or partner occupations were not recorded. Students were therefore recorded as a separate category.

The results demonstrate that people with a diagnosis of ARM are as likely as the general population to be of socioeconomic class I and II (see table 4). Therefore having a diagnosis of ARM does not affect one's ability to maintain a professional occupation. There were less ARM individuals recorded as having a manual occupation than the general public, this may be due to real or perceived physical limitations of their illness.

Table 4 –Summary of Socioeconomic Class (SEC)

Summary of the SEC of the participants compared with those of the British population.

SEC	n=	Percentage	Normal Population (150)
I	4	5.26%	3.8%
II	16	21.05%	22.1%
III non-manual	14	18.42%	28.9%
III manual	11	14.47%	20.6%
IV	6	7.89%	16.2%
V	10	13.16%	8.4%
Students	14	18.42%	No figures
Housewife	1	1.32%	No figures

In addition to employment questions pertaining to home and car ownership were also included. 46 of the 75 (61.3%) described themselves as being either home owners or

residing in a house owned by a family member. 47/75 (62.7%) owned one or more cars and 26 (34%) owned both a home and a car. Whilst this information is not directly related to their underlying condition it may be useful in interpreting clinical data where SEC may be important.

Also included in the demographic data collected was information regarding relationships and dependents. 46 respondents (61.3%) described themselves as being in a relationship, this was further categorised as; being in a relationship not living together, living together and married. Of those in a relationship 14 were married, 16 were living with their partner and 16 were not living together. Only 11(14.5%) had children, although 3 others were waiting to commence IVF treatment.

Clinical Data

ARM classification

Medical records were analysed for data on the original diagnosis. This was not possible in 11 individuals where medical records had been lost. The categories utilised in this study are low, high and cloaca. This is an oversimplification of other classifications that are in use, but seemed most appropriate given the retrospective nature of this part of the study. Therefore 26 of the 75 patients were considered to have a high anorectal malformation, 23 a low malformation, 15 patients with a cloaca (see table 5). The sub-division of patients into these groups is imperative to allow an accurate assessment of data and will permit cross analysis with previously published data.

Table 5 - Summary of diagnosis

Table showing the distribution of the level of anomaly from males and females.

	Number	Male n=31	Female n=44
Low	23 (31%)	6 (19%)	17 (39%)
High	26 (35%)	17 (55%)	9 (20%)
Cloaca	15 (20%)	0 (0%)	15 (34%)
No diagnosis	11 (14%)	8 (26%)	3 (7%)

Further details were also collected pertaining to the participants' initial surgical treatment. A colostomy is required in patients with high anomalies or cloaca prior to undergoing definitive surgical repair. The total number of colostomies performed in the immediate neonatal period was 39. Four of these were in the low ARM group, 21 of the 26 high ARM underwent colostomy formation and 14 of the 15 (93%) of the cloaca patients had colostomies. The majority of these were right transverse colostomies, but sigmoid loop colostomies and left descending colostomies were also performed. In 13 patients the exact type of colostomy procedure was not documented. One patient underwent total colectomy and permanent ileostomy formation as a neonate as a consequence of ischemic bowel.

All patients underwent definitive surgical treatment. The type of surgical procedure is dependent upon the level of the ARM and when the repair was undertake. Those individuals with a high ARM or cloaca born after 1984 are likely to have undergone a Pena repair. The initial surgical repair will be referred to as the index procedure, as subsequent repairs were necessary in some individuals. A summary of the types of procedures carried out can be found in table 6.

Table 6 – Surgical correction of ARM – initial procedures

Surgical Procedure	Number of Patients
Anal Cutback	20 (27%)
Abdominoperineal Pullthrough	17 (23%)
Rectal Pullthrough	9 (12%)
PSARP	7 (9%)
Sacroabdominal perineal Pullthrough	3 (4%)
Anoplasty	3 (4%)
Anal transposition	1 (1%)
Colectomy and permanent ileostomy	1 (1%)
No details available	14 (19%)

In addition to the initial procedure a further 39 surgical repairs were carried out on 31 participants, (this did not include treatment of rectal prolapse or minor anorectal

stenosis). The majority of these procedures were aimed at maintaining social continence as only 2 resulted in permanent bowel stomas. The rest were either re-do anoplasty, PSARP, anal transposition and abdominoperineal pullthrough. This does not include all procedures that may have been carried out in adolescence and adulthood at other institutions. Where possible medical records from hospitals other than the study institutions were sought, but not always successfully.

In addition to the initial repair this group underwent a significant number of surgical procedures as children. The average number of surgical procedures each underwent was 11 (range 0 to 77), all procedures performed under a general anaesthetic were included in this figure. Interestingly in the low ARM group the average number of surgical procedures performed for each individual was only 3.5 (SD =2.4), in the high ARM the average number of operative procedures was 12.67 (SD = 5.68) and perhaps not surprisingly this figure rose to 23 in the cloaca group (SD =9.55).

Associated Anomalies

Some of the increase in the number of surgical procedures may be accounted for by the increasing complexity of the problem requiring correction.

In addition to the anorectal malformation there are a significant number of other associated anomalies which may affect these individuals. Some of these may also require surgical intervention. The commonest associated anomalies in this group of affected individuals are those of spine and genitourinary tract. A summary of the associated anomalies is included in table 7.

Table 7 – Numbers of associated anomalies

A summary of the associated anomalies seen, the results have been given based upon the level of the original diagnosis.

	Low ARM n = 23	High ARM n = 26	Cloaca n=15	Total n = 64
Vertebral	7 (30%)	14 (54%)	7 (47%)	28 (44%)
Renal	4(17%)	6 (23%)	7 (47%)	17 (27%)
Urinary	6 (26%)	12 (46%)	8 (53%)	26 (41%)
Gastrointestinal	5 (22%)	6 (23%)	4 (27%)	15 (23%)
Genital	1 (4%)	12 (46%)	11 (73%)	24 (38%)
Cardiac	3 (13%)	5 (19%)	4 (27%)	12 (19%)
No Anomalies	9 (39%)	9 (35%)	0 (0%)	18 (28%)

Other anomalies were also documented in individual patients; in particular those affecting limbs were fairly prevalent; such as congenital dislocation of the hips, accessory digits and talipes equino varus. The presence of a further anomaly in addition to the anorectal malformation appeared to be a predictor for the occurrence others. In some patients the anomalies present may constitute a VACTERL association. This term is applied to individuals with three of more anomalies of the vertebral, anal, cardiac, tracheal, oesophageal, renal systems and radial limb defects (151). In our group of subjects there were 12/49 (24.5%) who were considered to have a diagnosis of VACTERL. Following accepted criteria, those with a diagnosis of cloaca were not included in these figures. Previous reports have put this figure at 15% (152).

Discussion

Demographics

It is reassuring to note that the demographic data of the participants of this study are broadly similar to that of the British population. It demonstrates that as far as possible, this is a representative group of individuals with a diagnosis of ARM and not a selected group. Furthermore, it provided useful and detailed data with regards to

the relationship status of the participants which may be useful in analysing some of the later results.

Clinical Data

Medical records were not available in 15% (11/75) of those that participated, despite considerable efforts by medical records department of Great Ormond Street Hospital and the researcher involved. Where paediatric and adult medical records were available the correlation between the two was often poor, with much of the information regarding surgery undertaken in childhood missing or inaccurate. In all cases there was no obvious reason for the lack of medical records, simply that they had been mislaid. As the participants were children they were dependant upon parents recollections, one individual did not realise that he even had a diagnosis of ARM, as he had an associated TOF which required urgent surgery, and was the only diagnosis that his parents could recall.

In addition it is possible that some of the participants underwent surgical treatment in establishments other than those involved in the study. Data recalled from this was often poor due to participants being unable to accurately remember where they had been treated as children and the difficulty in obtaining medical records from other hospitals.

Associated anomalies were seen in the majority of patients with ARM, and 100% of females with a diagnosis of cloaca. This is higher than the levels reported in some of the literature (153). In particular significant rates of anomalies were detected in those with low ARM. Some of the documented anomalies were minor spinal defects some of which were detected incidentally in adulthood as a result of investigation for other problems such as mechanical back pain.

Conclusions

This group of individuals' appear representative of the general population and the diagnosis of ARM does not seem to confer any negative impact on social status. Furthermore the majority of the participants were in a relationship, or had been in a

relationship. The majority owned their own car and home. These are all positive findings which should be presented to parents of affected children in order to allay fears over future outcomes for their offspring.

However, it should be noted that 15% of medical records were not available despite considerable efforts to find them. More stringent checks and audit of medical record departments should be recommended.

Chapter 5 - Bowel Function

Introduction

One of the principal aims of surgery for the repair of ARM is to establish bowel control and faecal continence. The degree of faecal incontinence is generally worse in patients with high ARM and cloaca. In addition to the poor development of the anal sphincter that these patients have, there are also a significant number with associated anomalies of the spine which may compound the problem.

The mode of defecation can be thought of as being *per rectum* or via a stoma. A significant proportion of the patients will defaecate per rectum either spontaneously or with some form of assistance; medication, enemas or rectal washouts. In cases where there is failure to achieve continence the patient may be offered a permanent colostomy, or an antegrade continence enema (ACE) procedure (154). This is used to avoid chronic constipation and the overflow incontinence that ensues, and gives the patient a degree of autonomy in their management. This procedure is of particular benefit to those who have impaired mobility and are unable to administer rectal washouts.

Abnormal bowel function can be clinically assessed as falling into two broad categories namely; constipation and incontinence. Both of these outcomes measures were assessed in this study using the Cleveland Clinic scoring systems (155). As previously published data on patients with ARM highlights, many of these patients are documented to suffer from a considerable degree of constipation. Pena proposes the underlying cause of constipation to be "...a hypomotility disorder of the rectosigmoid", this allows the sigmoid to distend and overflow incontinence (156).

Results

Mode of Defecation

Encouragingly the majority of the 75 participants were able to defecate spontaneously per rectum. Only 7 participants required regular oral medication or suppositories to

manage their bowel function. 18 had some form of stoma, the majority of whom had an ACE (see table 8).

Table 8 – Summary of Mode of Defaecation

Mode of Defecation	Numbers
Spontaneous per rectum	44 (59%)
Antegrade Continence Enema	11 (15%)
Permanent Stoma	7 (9%)
Rectal Washout	6 (8%)
Regular Laxatives	5 (7%)
Constipating Medication	1 (1%)
Regular Rectal Suppositories	1 (1%)

Cleveland Clinic Incontinence Score - CCIS

Of the 75 participants 8 did not respond to this section of the questionnaire; 7 of these had permanent stomas and one individual had an ACE. It is possible that the patients with stomas did not see these questions as relevant, as they related to the leakage of stool and flatus.

The CCIS (see appendices 3 & 4 for questionnaires) has a scoring range of 0-21, with 0 representing the best possible outcome of perfect continence and 21 being completely incontinent. Of the 67 respondents only 7 (10.5%) considered themselves to have perfect continence, although it is unclear whether the non-responders would also have considered themselves to have perfect continence, albeit with a stoma. 31 of the 67 (46.3%) scored between 1 and 7, which is interpreted as 'good continence'. The range within this group is fairly wide, as passing flatus unintentionally places an individual in the same category as an individual who occasionally is incontinent of solid stool. It is likely that these two situations could not be considered similar in the level of embarrassment and distress that they would cause the individual.

21 (31.3%) patients described themselves as having moderate incontinence; this represented a score of between 8 and 14. Furthermore 8 subjects (12%) scored between 15 and 20 points, which is considered to be severe incontinence. No

participant scored a full 21, which represents the category of complete incontinence. See table 9 for a summary of results.

As expected the level of incontinence seen was dependent upon the level of malformation the patient was born with. Those with a diagnosis of low ARM had a mean CCIS of 5.43 (SD=5.45), representing good continence. Patients with a high ARM had a mean score of 8.13 (SD=6.47), and those with a diagnosis of cloaca had the highest average incontinence score with a mean of 9.64 (SD=5.74).

Table 9 – Summary of Cleveland Clinic Incontinence Scores and level of ARM

	Score = 0 Perfect	Score 1- 7 Good	Score 8 – 14 Moderate Incontinence	15 -20 Severe Incontinence	No answer
Low ARM (n=23)	6 26%	11 48%	4 17%	2 9%	0 0%
High ARM (n=26)	0 0%	12 46%	7 27%	4 15%	3 12%
Cloaca (n=15)	1 7%	2 13%	7 47%	1 7%	4 27%
No Available Diagnosis (n=11)	0 0%	6 55%	2 18%	2 18%	1 9%

CCIS Scores in patients requiring assistance.

In those participants who required assistance with defecation it may be anticipated that they would have poorer CCIS scores. However this did not seem to be the case. Whilst there were low levels of perfect continence the levels of moderate to severe incontinence were not significantly higher than those seen in the group as a whole (see table 10).

Table 10–Cleveland Clinic Incontinence Scores in patients requiring assistance with defecation
Incontinence scores for those individuals who do not defecate per rectum

	Did not answer	Perfect	Good Continence	Moderate Incontinence	Severe Incontinence
Stoma n=7	6 (86%)	0 (0%)	1 (14%)	0 (0%)	0 (0%)
ACE n=11	2 (18%)	1 (9%)	1 (9%)	4 (36%)	3 (27%)
Rectal Washout n=6	0 (0%)	0 (0%)	4 (67%)	1 (17%)	1 (17%)
Total (n=24)	8 (33%)	1 (4%)	6 (25%)	5 (19%)	4 (15%)

Associated anomalies

In addition to the original anomaly it is probable that associated anomalies, particularly spinal anomalies, may also have an effect on faecal continence. Twenty five of the participants had documented spinal anomalies, the majority of these were sacral. In the group of patients with good or perfect continence 8 of 38 (21%) had sacral anomalies, in those with impaired continence 17 of 27 (59%) had documented sacral anomalies. The remaining 8 patients did not answer the CCIC questions, as mentioned previously, but 3 of these did have spinal anomalies.

Type of repair

The possibility that bowel function after ARM is dependent upon the type of initial repair performed has been mentioned previously. The majority of our subjects underwent a pull-through or cut-back procedures as their initial repair. Only 7 individuals had a PSARP operation performed initially (see table 7 for summary of initial repairs). This procedure was not widely practiced in the UK until recently and this study only looks at adult functional outcomes.

With regards to bowel function, of those who had undergone a pull-through procedure as their initial repair 3 were left with permanent stomas. 13/29 (45%) described themselves as having good continence. 8/29 (28%) had moderate incontinence and a further 5/29 (17%) had severe incontinence. 3 individual did not answer this section, 2 of these had permanent stomas and presumably did not think the questions relevant.

Those individuals who had undergone a PSARP 4/7 (57%) had either perfect or good continence. The remaining 3 (43%) had moderate incontinence. Therefore, there was no significant difference between those who had undergone the pull-through procedure, however, the numbers who underwent a PSARP as their initial repair are small. It should be noted that none of those who had undergone a PSARP repair required a subsequent stoma to manage their bowel function. Furthermore, those individuals who had undergone a Pull-through procedure were generally older than those who had undergone a PSARP. The average age of the Pull-through group was 27.15 years compared with 20.71 years in the PSARP group ($p=0.03$). It may be that those who had undergone a PSARP maintain improved continence into adult life and subsequent studies may demonstrate this.

The mean age of sexual debut for the group was 17.6 years and this was not influenced by degree of continence. In those with perfect or good levels of bowel continence the mean age of sexual debut was recorded as being 17.7 years. In those with moderate incontinence it was 16.7 years, and in the group with poor continence this raised to 18.3 years. As discussed previously, this is slightly later than the UK average age for first intercourse.

Constipation Scoring System

Constipation is a symptom that is difficult to assess, as the pattern of normal bowel habit can vary from once every 3 days to 2-3 times per day. Infrequency may be due to many factors, many of which are due to individual factors including diet and exercise. The Constipation Score (see appendices 3 & 4 for questionnaires) covered variables including; frequency of bowel movements, painful evacuation, incomplete evacuation, abdominal pain and duration of constipation. Each variable is scored and an overall score of greater than 15 is used to define constipation.

Only 3 (4%) of our subjects scored greater than 15 and could be classified as constipated. The mean score was 7.8 (SD=5.1). Of the 3 that scored sufficiently high to be described as constipated, 2 had undergone the ACE procedure for a persistent cloaca, the other individual had a low ARM with a scoliosis and short sacrum. In addition 5 (6.67%) subjects required regular laxatives, it is likely that they may have scored more highly if they were not taking any medication. Overall, only 12% of participants were constipated.

Quality of Life

The concept that incontinence and stomas may have an adverse affect on quality of life is well documented (157). In order to assess the impact of having a stoma, an ACE and having to perform rectal washouts, on quality of life, the results of the SF36 questionnaire were analysed for these individuals. Generally, those with an ACE tended to fare worse than those with permanent stomas or those performing rectal washouts, although the numbers involved are too small for any meaningful statistical analysis (see table 11).

Table 11 – SF 36 mean scores over all categories

Quality of life scores for those individuals who do not defecate per rectum.

Variable	Stoma (n=7)	ACE (n=11)	Rectal Washout (n=6)
Physical Functioning	78.57	80.45	95
Physical Role	85.71	77.27	95.83
Bodily Pain	79.36	77.77	87.03
General Health	59.71	57	60.58
Vitality	72.85	58.18	70.85
Social Functioning	88.88	75.75	90.74
Emotional Role	90.47	78.78	66.67
Mental Health	77.71	67.91	69.33
Change in Health	57.15	61.36	50

Further analysis of the quality of life scores was undertaken, including comparison between those with a low and high ARM and no statistical differences were noted. More detailed analysis of quality of life and comparison of normal values data are included in chapter 8.

Discussion

Incontinence

Overall the results were optimistic with the majority (57%) describing themselves as having either perfect or good continence. Nevertheless, it is apparent that there are a significant number of individuals who had symptoms of faecal incontinence. It should be noted that these results did not include the 7 individuals with permanent stomas. Whilst these individuals are “socially” continent, having a stoma cannot be considered a good outcome.

Furthermore, it is encouraging to note that those individuals suffering from considerable faecal incontinence are sexually active, and that the mean age of sexual debut was not that much later than those individuals with good/perfect continence.

Low ARM

74% of our subjects with a diagnosis of low ARM had good continence, this is comparable to other reports in the literature (158-160) where the range of faecal continence is reported to be from 60-80%. Very few individuals (6/23) described their continence as perfect. In addition to this Rintala et al reported the presence of sexual dysfunction in 13%, and social functioning problems in 39%. This level of sexual and psychological difficulty was not seen in our cohort.

High ARM

Almost half of those with a diagnosis of high ARM described themselves as having good faecal continence. The rest had moderate to poor incontinence or permanent stomas. Again this is largely concurrent with the published literature on adult bowel

function after ARM repair (161;162). Other studies have reported better outcomes (163) with over 60% having good/fair bowel function and 15% normal function. The reason behind these differences is not obvious, and may be a result of the method of data collection, individual surgical technique or bowel management facilities and support.

Significant levels of social disability have been previously described in this group of individuals. Rintala et al (164) report that in their series 85% of adults complained of some form of social limitation as a result of faecal soiling. These results were not replicated in our individuals where there were no statistical differences noted in the SF-36 subscales between those with a high or low ARM.

Constipation

The rate of constipation seen in this study are much lower than those published in the literature, which vary from 22 -62% (40;165;166). However there is evidence to suggest that constipation is a symptom that children grow out of (167), although unlike this study all of the subjects in this review had undergone the PSARP procedure. Other possible explanations for this include the method by which constipation is assessed. The scoring system utilised in this study was devised for patients in the USA with idiopathic constipation, those with mechanical or neurogenic aetiologies were excluded (168). It may be that this was not an ideal scoring system to use in patients with a diagnosis of ARM. A 7 day bowel diary may have provided more useful and detailed data. However, it is likely that compliance with this would have been poor. Nevertheless it is clear that attempting to quantify what is a particularly subjective symptom is always going to be difficult. This is in contrast to incontinence where the number of episodes can be quantified with greater ease.

Only 7 individuals had undergone a PSARP as an initial procedure, none of whom considered themselves to be constipated, 3 had gone on to have further surgery to improve their bowel function, in 1 case this was a re-do PSARP and 2 others required a ACE. Eight subjects underwent a PSARP as a secondary repair after failure of their initial repair. Of these 2 required regular rectal washouts to eliminate, and 2 had a subsequent ACE. Whilst none of these scored a score consistent with being

constipated it may be that those requiring regular washouts would be constipated if they did not perform rectal washouts.

Other studies looking at bowel function outcomes after ARM repair do not use constipation scores, but consider those that require regular enemas, rectal washouts or laxatives to be constipated. However, it has been previously acknowledged that these treatment options can be offered on an indiscriminate basis (79) and therefore it may be postulated, should probably not be used as means of quantifying constipation. It may be that in future assessments of ARM that subjective symptoms such as constipation should not be used as a measure of outcome. From the authors own experience with this group of patients, constipation was not a symptom that many patients reported to be particularly bothersome.

Type of Initial Repair

As demonstrated above this does not seem to be the only explanation for the difference seen in incontinence rates. Bowel function may be impaired by the type of operation, as more of those who had a pull-through ended up with incontinence and stomas compared with the PSARP. However, perhaps a caveat to this fact should be that as yet we have no information on bowel function of those who have undergone a PSARP when they are of a comparable age.

ACE

In total 13 of our respondents had undergone an ACE procedure. Eleven were actively using them, one individual had gone on to have a colostomy and one had undergone a reversal and was using regular oral laxatives. Although the reasons for this were not documented, presumably this was due to failure to manage their bowel function adequately with the ACE. Only 2 of the 11 (18%) reported having either perfect or good continence, which is the aim of performing this procedure. It is possible that the patient with an ACE that did not respond to the CCIS questions did not experience any adverse bowel symptoms as a consequence of his ACE

Furthermore, when quality of life scores were analysed it was noted that those individuals who had undergone an ACE tended to score more poorly than those individuals who either performed regular rectal washouts or had a permanent stoma (see table 11). This was an unexpected finding and has not been reported before. The general perception is that an ACE improves quality of life and anecdotally those individuals who had an ACE reported an improvement in quality of life and ability to manage their bowels when interviewed by the main researcher.

It is possible that whilst the ACE procedure is appropriate in children and adolescents, rapid growth during puberty may have a deleterious effect. One possible solution to this problems is to consider that use of an ACE in combination with a neosphincter (169). Whilst this by no means guarantees faecal continence it may be an option in those contemplating permanent colostomy.

Conclusions

Overall, our results for faecal incontinence are comparable to those published in the wider medical literature. For a more meaningful interpretation of these results they should not only be considered in terms of the original diagnosis but also the surgical repair, or repairs that were required to achieve this result. However given the retrospective nature of this type of data collection despite best intentions this is likely to be very unreliable evidence. Furthermore, very few of the published data include details of re-operation rates and specifically their effects on bowel function.

Of those individuals who were incontinent none had undergone any form of artificial or gracilis sphincter repair. Whether this is because the majority were not routinely under follow-up with a colorectal surgeon is not clear. The benefits of such treatments have been documented not only in terms of improved continence but also in terms of quality of life (170).

The results regarding sexual satisfaction and body image demonstrate that despite considerable morbidity associated with diagnosis, treatments and associated anomalies, the majority of our participants are functioning well both physically and

psychologically. As this is the largest series of ARM individuals in the UK, where this aspect of functioning has been fully assessed this aspect should be highlighted to parents with affected children.

Chapter 6 - Urinary Function

Introduction

The impact of ARM on urinary function comes not only from the abnormal fistulous communication between the anus or rectum with the urethra or bladder, but also from any associated urinary tract congenital anomalies and potential iatrogenic injury to the urinary tract that may occur during surgical repair. One of the key aims of ARM repair is to maintain renal function. A persistent fistulous communication between the bowel and urinary tract predisposes to infection and prior to modern paediatric surgical technique was a major cause of morbidity and mortality in this population.

In order to assess urinary function participants were required to answer simple questions with regards to their method of micturition. This was considered to fall into one of the following categories:

- Voids spontaneously
- Clean Intermittent self catheterisation (CISC)
- Continent Diversion e.g. Vesicostomy
- Incontinent Diversion e.g. Ileal conduit

In addition to these questions the Bristol Female Lower Urinary Tract Symptoms Questionnaire (BFLUTS) (127) was used to assess lower urinary tract symptoms (see Chapter 3 for more information). The BFLUTS is a well utilised questionnaire which has been widely used in other studies and validated on a UK population. The male urinary function questionnaire developed by the same group is the International Continence Society (ICS) male questionnaire (171). This has also been validated on a UK population.

Results

GU Anomalies

Urinary tract anomalies were documented in 35/66 (53%). In the cases of 9 participants no medical records were available to accurately assess underlying diagnoses. Anomalies of the reproductive system were not included as these will be discussed in more detail in chapter 7. Of those affected 7 had a documented low ARM, this represented 30.43% of the entire low ARM group. This percentage is comparable to other published data from the same centre (172). 14 had an initial diagnosis of high ARM and 53.85% of this group had an associated anomaly. 12 females (80%) with a cloaca were affected. 2 of the affected individuals did not have an initial diagnosis as their medical records were not retrievable. In total 50 anomalies were recorded (see table 12).

Table 12 - Summary of Urinary Tract Anomalies

Anomaly	Number of Patients
Vesicoureteric Reflux	23 (35%)
Solitary Kidney	7 (11%)
Dysplastic Kidney	4 (6%)
Atonic/Neuropathic Bladder	5 (8%)
Urethral Diverticula	3 (5%)
Duplex	2 (3%)
Ectopic Kidney	2 (3%)
Multicystic kidney	2 (3%)
Malrotated Kidney	1 (1.5%)
Patent Urachus	1 (1.5%)
Left PUJ Obstruction	1 (1.5%)
Ureterocoele	1 (1.5%)
Congenital small bladder	1 (1.5%)

The commonest anomaly detected was vesicoureteric reflux. In the majority of cases this was diagnosed and treated if necessary. Only 6 of the 23 individuals with reflux underwent any surgical intervention for reflux. Four underwent ureteric re-

implantation, one individual underwent 2 STING (Subureteric Teflon injection) procedures and 1 had a transureteral ureterostomy. In one case the diagnosis was not made until adolescence and a nephrectomy was required to remove the affected non-functioning kidney. In addition 3 male participants had undergone renal transplantation, 2 of whom had live donors, one patient received his donated kidney from his wife. Furthermore 3 patients were in chronic renal failure, one of whom was on the waiting list for a renal transplant.

With regards to bladder reconstruction, 2 individuals required bladder-neck reconstruction. Three required bladder augmentations, in all cases ileum was used. In those with augmented bladders 1 had problems with bladder stones, as did the female patient who relied upon manual expression to empty her bladder. In one male with a neuropathic bladder managed with CISC, recurrent urinary tract infections resulting in epididymitis necessitating a vasectomy to relieve the problem.

Mode of Voiding

The mode of voiding is an important aspect of day to day living for patients with ARM, and can have a significant impact upon an individual's quality of life. The majority of participants (58/75) are able to spontaneously void (see Table 13). Only 8 had some form of stoma, the majority of which were continent diversions. All but one of the individuals with a Mitrofanoff are females with a diagnosis of cloaca.

Table 13 – Methods of Bladder Emptying

Mode of Voiding	Males	Females
Spontaneous Voiding	26 (34.7%)	31 (41.3%)
CISC per urethra	3 (4%)	6 (8%)
Mitrofanoff	1 (1.3%)	6 (8%)
Ileal Conduit	1 (1.3%)	0 (0%)
Expresses Bladder	0 (0%)	1 (1.3%)

The majority were able to void spontaneously, only one individual had an incontinent diversion, his case was complicated by the fact that he had required a renal transplant,

as a consequence of reflux in a solitary kidney. In those that performed CISC no problems were reported.

Incontinence

Urinary incontinence is defined as the involuntary passage of urine at a socially inconvenient time. Incontinence was assessed using the self-reported questionnaires. There were also questions regarding the frequency of any incontinence and whether pads were required. No quantitative measures of the degree of incontinence such as pad testing were employed.

Thirty of the participants (40%) reported that they were incontinent of urine to some degree (see Table 14). The majority of those that were incontinent were wet only on a weekly basis. Only 13 individuals described themselves as being incontinent on a daily basis, 4 of these stated that they were wet all of the time. Ten of these 13 required incontinence pads on a daily basis, a further 10 individuals required incontinence pads occasionally.

The BFLUTS/ISC questionnaires are able to discern between stress, urge, nocturnal and unexplained incontinence. The commonest pattern of incontinence was a mixed picture with 12 individuals affected. Only 5/75 complained of only stress incontinence symptoms. 4/75 reported urge incontinence and 4 had nocturnal incontinence. No patients were taking medication for their incontinence.

Table 14– Summary of rates of urinary incontinence

Table demonstrating the frequency of urinary incontinence in the different diagnosis categories.

Degree of incontinence	No notes available n=11	Low ARM n=23	High ARM n=26	Cloaca n=15	Total n=75
All of the time	0 (0%)	0 (0%)	1 (3.9%)	3 (20%)	4 (5.3%)
Daily	1 (6.7%)	1 (4.4%)	3 (11.5%)	4 (26.7%)	9 (12%)
Weekly	1 (6.7%)	8 (34.8%)	4 (15.3%)	4 (26.7%)	17 (22.7%)
Monthly	0 (0%)	0 (0%)	0 (4.4%)	1 (6.7%)	1 (1.3%)
None	9	14 (60.9%)	18 (69.2%)	3 (20%)	44 (58.7%)

Previous studies have suggested that the presence of incontinence may have a negative impact on the ability to form and maintain personal relationships (173). The results were analysed to see if there was a relationship between incontinence and the method of micturition and the ability to form personal relationships. Using a 2-tailed t-test the degree of incontinence was assessed as a variable against the participants stated relationship status, no significant difference was found between those that were in a relationship and those that were not ($p=0.53$). A similar result was found when the method of micturition was looked at, those that either performed CISC or had some form of diversion were as likely to be in a relationship as those that voided spontaneously ($p=0.548$). Interestingly, one variable that seemed to have an impact on relationship status was socioeconomic class, with those in SEC 1 or 2 were significantly more likely to be in a relationship than those in SEC 3-5 ($p=0.004$).

Lower Urinary Tract Symptoms (LUTS)

The relationship between ARM and LUTS is often overlooked in favour of assessing urinary incontinence. However the impact of LUTS particularly on quality of life is an important issue which has been addressed by several large studies recently (174;175). The majority of participants did not experience a significant degree of

LUTS which is reassuring; however rates were higher in the females (see table 15). Of those symptoms listed in the table below, urgency and incomplete emptying were reported as being the most bothersome. Those that failed to answer this section of the questionnaire included those dependent upon CISC either per urethra or via a Mitrofanoff.

Table 15 – Frequency of LUTS

Lower urinary tract symptoms as assessed by the ICS/BFLUTS questionnaires.

Symptom	Females n =44	Males n =31
Frequency	12 (27%)	4 (13%)
Nocturia	8 (18%)	2 (6%)
Urgency	25 (57%)	10 (32%)
Dysuria	19 (43%)	6 (19%)
Incomplete Emptying	22 (50%)	15 (48%)

Sixteen of the 75 (21.3%) complained of frequency (voiding at least every 2 hours), 12 of those affected were females. Only 6 (37.5%) of those affected found this to be a bothersome symptom. Nocturia was reported in 10 (13.33%) individuals, those that voided on one occasion during a night were not included. All of those complaining of nocturia reported it to be a bothersome symptom; one participant reported voiding 4 times per night or more.

Perhaps unsurprisingly, the frequency of LUTS seen in the participating females is higher than those reported in normal control data which is derived from females with no clinical problem (127). However when a comparison is made between ARM females and females with a diagnosis of Congenital Adrenal Hyperplasia (CAH) we can see that the results are broadly comparable (see table 16). The data collection method for the ARM females and CAH females is identical and was undertaken by the same group of researchers.

Table 16 – Comparison of LUTS in Females with ARM, CAH and Normal controls.
A Summary of results of the BFLUTS questionnaire in ARM females, normal controls and females with CAH.

	ARM females n=44	Normal Females (127) n=20	CAH Females (176) n=19
Frequency	26%	0%	21%
Urgency	58%	20%	68%
Nocturia	18%	5%	21%
Dysuria	43%	15%	53%

Discussion

Levels of urinary tract anomalies were high with approximately half of the participants being affected. This figure is similar to previously published literature (177;178) which puts the rate of associated urinary tract anomalies at about 40%. In addition 35% had vesicoureteric reflux, which again is a figure comparable to the published literature (179). However as not all participants had undergone investigation with a MCUG it may be that the correct figure is actually higher than this.

Associated Anomalies

Long-term consequences of reflux were satisfactory in the majority of patients. There were 2 cases of transplant directly related to reflux, one in the case of late diagnosis and a further in an individual with a solitary kidney. A further 2 individuals had chronic renal failure secondary to reflux in solitary functioning kidneys and 1 individual was hypertensive.

Incontinence

The degree of urinary incontinence appeared to be higher than previously published. The largest study looking at degrees of urinary incontinence in this group was by Pena and included children and adults. Pena found no incontinence in his patients with

corrected low anomalies. Whilst the rates of incontinence in this study are also lower in the low ARM group with only 1 person complaining of daily incontinence, a third of individuals had incontinence on a weekly basis. The difference in the reported rates may be explained by the method of data collection. Questionnaires may encourage people to report even minor symptoms that are not perceived as being bothersome.

In those with a diagnosis of high ARM the levels of incontinence were also higher than previously quoted. 15% described themselves as having episodes of incontinence on a daily basis, this figure is three times higher than the 5% quoted by Pena in his series (180). One of the possible explanations for this is that our series consists of only adult patients and urinary incontinence has developed as the patients have aged. In addition our patients were operated upon by more than one surgeon. Therefore there may be an operator bias. In addition these are self-reported symptoms rather than acquired in an interview style which is how much of the published data in the literature is acquired. This style of information acquisition has been previously demonstrated to increase rates of patient symptoms (181). Nevertheless, even with a high ARM the majority were completely continent of urine.

Unfortunately the majority of participants with a cloaca reported urinary incontinence with only 3/15 reporting that they were completely dry. For 40% incontinence was on a daily basis. This fact is more problematic considering that 7 of these had undergone a 'continent' diversion which in the cases of 6 did not prevent incontinent episodes. Furthermore, 4/7 (57%) were incontinent on a daily basis. This should be considered when counselling patients for major reconstructive surgery as making the affected individual dry may simply be impossible. Just over a quarter of the females with a cloaca were dry, and this figure includes those that reported incontinence on a monthly basis as this was not considered to be a significant problem.

LUTS

Both male and females participants reported LUTS and although less common in men, both groups were bothered by these symptoms. The higher rates reported in the female participants is in part explained by the fact that the most severely affected individuals with the diagnosis of cloaca are all females. Where LUTS are present in

males they are as bothered by the symptoms as their female counterparts with a similar degree of symptoms. This study presents the first detailed data on the presence of LUTS in ARM.

Interestingly the prevalence of LUTS in females with ARM is fairly similar to females with CAH, despite the fact that the urinary tract is not the site of primary pathology. A possible explanation for this includes pelvic surgery in childhood as this would seem to be the only common factor between these two groups.

As for incontinence, these symptoms were more common in our study participants and this may be due to our method of assessment. Rodriguez et al found a significant difference in the reported levels of a patient's urinary symptoms when assessed by a doctor-led interview and a patient completed questionnaire (142), with symptoms consistently underestimated in the doctor-led interview. This may be due to reluctance of patients to express dissatisfaction to doctors or embarrassment in discussing symptoms of a personal nature.

Neuropathic Bladder

The incidence of neurogenic bladder in our series was only 6%, including cases that were secondary to spinal anomalies or iatrogenic injury. This diagnosis was recorded if it had been clearly documented in the medical records. In addition if participants were under regular urological follow-up or practiced ICSC then the possibility of neuropathic bladder was considered. This study demonstrates a rate lower than previously stated in the literature (182), although it should be noted that this study looked at a population specifically attending a urology clinic after anorectal repair, and therefore is heavily biased in favour of those patients with specific urological problems. Other studies put this figure closer to the 10% (183) to 18% (184). There are data to suggest that iatrogenic neuropathic bladder is minimised with a PSARP approach (185). In this study the numbers were too small to analyse the patients who had undergone different surgical procedures.

The consequences of CISC are generally considered minimal with the risk of urinary tract infection, however the possibility of squamous metaplasia and carcinoma particularly in those with spinal anomalies should be considered.

Conclusions

The degree of renal anomalies seen in our group is comparable to other studies. The majority of patients void spontaneously and are dry. Unfortunately those who had undergone reconstruction did have significant levels of incontinence which has not been previously reported in the literature. Possible explanations for this are that the surgery was in many cases performed many years previously and this is the long-term sequelae of bladder reconstruction and diversion in adolescents and adults.

There is evidence to suggest that those individuals with an abnormal bladder in childhood who undergo surgery can expect a deterioration in adolescence (186) and it may be that when other groups look specifically at their adult outcome data that a similar picture may emerge. In addition, this study involved clinicians not directly involved in the participants' current or previous medical care, which may have eliminated any reporting bias and gives a more objective view of long-term outcomes. External audit of surgical outcomes should be undertaken in units to allow an objective comparison to be made.

There are those that would argue that more modern surgical techniques minimises the risk of subsequent incontinence. Although this has not been proved in any studies comparing directly adult patients who have undergone one type of repair with the PSARP.

Chapter 7 –Sexual Function & Fertility

Introduction

Sexual functioning and fertility in individuals with ARM may be impaired for a number of reasons;

- Associated malformations of the genital system
- Iatrogenic injury at the time of surgical repair either to genital/reproductive structures, or to the nerve supply of these structures
- Psychological problems as a consequence of ARM may impair the ability to form and maintain relationships
- Social isolation, which may be associated with urinary or faecal incontinence

Those who have normal sexual function may also experience difficulties with fertility. In male patients these two aspects may be inextricably linked, men with erectile difficulties may also experience difficulty with ejaculation. These individuals who are unable to ejaculate may require sperm extraction techniques to assist fertility. The extent of these problems in the adult ARM population is difficult to assess as many of the long-term outcome studies are carried out to include a paediatric population where questioning about sexual activity and fertility would be inappropriate.

In females where penetrative sexual intercourse is possible, and the Mullerian structures are normal, pregnancy is possible. Nevertheless, there are concerns about ARM patients in pregnancy. The potential damage that could occur during the course of pregnancy and childbirth include:

- possible deterioration in renal function – particularly if a degree of renal failure already exists
- worsening urinary incontinence, or damage to urinary diversions during delivery
- decreased faecal control as a result of pelvic floor injury

All participants completed questions regarding sexual debut and any pregnancies they or their partners may have had, and whether they resulted in a live birth or not. Furthermore they were also asked to complete an abbreviated version of the Snell Multidimensional Sexual function questionnaire (135). Male participants also completed the Brief Inventory of Sexual Function (187). Female participants were also questioned about menstruation. The details of these questionnaires have already been discussed in chapter 3 and are included in appendices 3 & 4.

Results

Of the 75 subjects, 10 were not sexually active (13.3%). The reasons for this were not always obvious, although 2 did mention that this was through personal choice. In the case of 2 females it was due to vaginal stenosis, both required further surgery after examination of the perineum revealed that there was no vagina suitable for penetrative sexual intercourse.

The mean age of sexual debut for the group was 17.65 years (range 14 – 65 years). The mean age of sexual debut in males and females was similar, 17.4 years in females and 18 years in males. Furthermore, there were no significant difference between the low, high and cloaca groups, whose mean age in years at sexual debut were 17.3, 17.8 and 17.6 respectively.

Fertility

41 of the 44 females reported that they menstruated normally. The 3 that did not had undergone hysterectomy. One as emergency treatment for uterine prolapse after a vaginal delivery, and the other 2 during surgery for repair of their cloacal anomalies. One patient was not aware that she had undergone a hysterectomy but had been informed that she was born without a uterus.

There were a total of 16 pregnancies from 7 females, 10 of which resulted in live births. The majority of these (5/7) had low ARM, one had a diagnosis of high ARM and in one female the diagnosis was not clear. Six of these were via Caesarean section and 4 vaginal deliveries. There were also 4 documented miscarriages and 2

terminations. In addition one participant had adopted 2 children as she had been told she would be unable to carry a pregnancy to full term, although no mullerian anomaly had been documented and she had normal periods and vagina. She did have spina bifida, but was mobile with walking aids. One female was on the waiting list for IVF treatment to commence as she had not been able to conceive naturally.

In the male participants there were 11 documented pregnancies in their partners resulting in 10 live births. Four men had required fertility assistance in the form of IVF and a further 2 were currently undergoing fertility assessment.

Genital Anomalies

Eighteen genital anomalies were recorded in 14 of the female patients (32%). For a summary of the types of anomaly found see table 17 below. In females with a genital anomaly all had a high ARM or cloaca, none of the low ARM individuals were affected.

Table 17 - Summary of Female Genital Anomalies

Anomaly	Number (n=44)
Double Vagina	6 (14%)
Bicornuate Uterus	5 (11%)
Uterus Didelphys	2 (5%)
Absent Vagina	1 (2%)
Double Cervix	1 (2%)
Labial Fusion	1 (2%)
Mullerian Anomaly – not specified	1 (2%)
Vaginal septum	1 (2%)

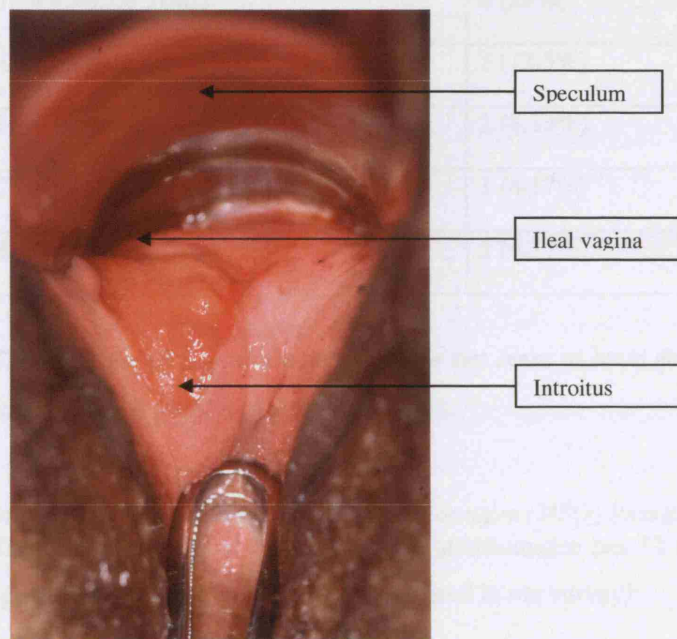
In those 14 females with a Mullerian anomaly all but 2 had anomalies of other systems; the other 12 individuals had other, often multiple anomalies.

In addition to their original surgical reconstructions, 4 females with a diagnosis of cloaca had undergone a further vaginoplasty, 2 of whom had also had a hysterectomy. One of these vaginoplasties was carried out using ileum (see figure 14), at the same time as a

neobladder reconstruction using ileum. In addition 4 others had vaginal stenosis, 2 were being treated conservatively with vaginal dilators and 2 required further surgical interventions.

Figure 14 - Ileal Vagina

Clinical photograph of a vagina constructed from ileum in a woman born with an anorectal malformation. The appearance is satisfactory, but persistent coital haemorrhage has been a severe impediment to regular intercourse.



In this study 10 of the 24 (41.67%) males where paediatric medical records were available had 11 documented genital anomalies. The commonest of these being cryptorchidism (see table 18). The majority of the males with genital anomalies (9/10) were sexually active.

Table 18 - Summary of Male Genital Anomalies

Anomaly	Number
Undescended Testis	6 (25%)
Hypospadias	3 (12.5%)
Bifid Scrotum	2 (8.33%)
Chordee	1 (4.17%)
Hydrocoele	1 (4.17%)

The presence of a genital anomaly does not seem to have an adverse effect on later sexual activity in both males and females.

Multidimensional sexual function questionnaire (MSQ) Results

The Multidimensional sexual function questionnaire has 12 subscales which look at (those highlighted are the subscales utilised in our survey):

- **Sexual esteem**
- Sexual preoccupation
- Internal sexual control
- Sexual consciousness
- Sexual motivation
- **Sexual anxiety**
- **Sexual assertiveness**
- **Sexual depression**
- External sexual control
- Sexual monitoring
- **Fear of sex**
- **Sexual satisfaction**

In an attempt to try and rationalise the length of survey that participants were required to complete an abbreviated version of this questionnaire was used. The original MSQ as described by Snell (135) contains 61 questions forming 12 subscales each of 5 items. Responses are recorded by the participant using a five point scale. Higher scores represent greater amounts of the respective sexual tendencies. Therefore a high score in the sexual satisfaction subscale would indicate a good result; however a high score in the sexual anxiety subscale would indicate a poor result. Due to considerations of length and applicability, only six of the subscales are used in this study; sexual esteem, sexual anxiety, sexual depression, sexual depression, fear of sexual relationships, sexual assertiveness and sexual satisfaction. This abbreviated version is already utilised in our research department with satisfactory outcomes. These surveys were completed by all but two of the participants, one of whom stated that he was not sexually active for personal reasons.

The results demonstrate that in two of the subscales; sexual assertiveness and sexual satisfaction there are significant differences between the males and females with ARM (see table 20). It would appear that females with a diagnosis of ARM are likely to be less sexually satisfied than their male counterparts, which is not true of the normal population upon which the questionnaire survey was validated on. Furthermore, they have significantly lower sexual esteem, and are therefore less likely to be able to relate with others on a sexual level (135).

Table 19– MSQ Results

Sexual esteem, sexual assertiveness and sexual satisfaction are positive subscales and a higher represents a better outcome. Sexual depression, fear of sex and sexual anxiety are negative subscales and higher scores represent a poor outcome. For all parameters the ARM group fares worse than normal controls.

Subscale	Males ARM	Males Normal	Females ARM	Females Normal	t-test comparing males & females ARM
Sexual Esteem	11.33	13.93	8.13	12.41	0.0157
Sexual Assertiveness	10.04	11.41	7.55	10.24	0.477
Sexual Satisfaction	11.37	12.53	10.00	12.95	0.0012
Sexual Depression	4.06	3.88	5.55	3.89	0.2892
Fear of Sex	7.78	6.53	7.65	8.63	0.86
Sexual Anxiety	6.25	5.68	7.35	5.25	0.3433

Brief Male Sexual Function Inventory Results

Only male patients completed this validated questionnaire. There are 5 sub-scales in this questionnaire looking at; sexual drive, erections, ejaculation, problem assessment and overall satisfaction. All but 1 of the males answered the questions. Five individuals (16.67%) described themselves as having poor erections or difficulty in getting an erection. The reason for the poor or absent erectile function was not examined in this study. Normal erection is dependent upon an intact reflex arc from the afferent stimuli to the glans penis via the dorsal nerve of the penis and pudendal nerve to the erection centre (S2-S4). The efferent fibres are distributed from the erection centre via the pelvic plexus to the deep artery of the penis. Any abnormality

or injury to this mechanism may impair erection. The possibility of a non-organic cause of erectile dysfunction is also possible, although not common.

Furthermore, 5 individuals reported that they were unable to ejaculate or had only a small amount of ejaculate. The mechanism of abnormal or absent ejaculation is more complicated. The possibility of retrograde ejaculation was also not addressed by the Brief Male Sexual Function Inventory, although none of the patients questioned described this as being a particular problem. Ejaculation is a more complex function dependent upon intact sympathetic and parasympathetic pathways, and an intact bladder neck.

Six participants (20%) reported a low sex drive. Sex drive dysfunction is difficult to interpret because of its multi-factorial nature. Only one of the participants was taking medication to improve his erections. Nevertheless, the majority of males 61%, described themselves as being very or mostly sexually satisfied (see Table 20). When compared to normal controls it is possible to conclude that the percentages of men very or mostly satisfied are comparable. However men with ARM are more likely to be dissatisfied. It should be noted that the control data was derived from an American population study where the youngest participant age range was 40-49 years which is the data used in table 20.

Table 20 – Male Sexual Satisfaction Scores

Using the sexual satisfaction subscale of the Brief Inventory males with ARM are compared to normal control males.

Score	Number of Males n=31	Normal Control Data (188)
Very/Mostly Dissatisfied	7(23%)	12.5%
Neutral	4 (13%)	27.1%
Very/Mostly Satisfied	19 (61%)	60.4%
No answer	1 (3%)	0%

Using the sexual satisfaction subscale to assess for any obvious variations within subgroups it became clear that factors previously postulated as having an adverse affect on sexual outcomes did not appear to change the sexual satisfaction score. Faecal incontinence or fear of faecal incontinence has been described as a possible factor in the adverse outcomes of these patients. Nine males described themselves as having a significant degree of faecal incontinence (in this study faecal incontinence is defined as a Cleveland Clinic Incontinence Score of 8 or more). This group of individuals did not have lower sexual satisfaction scores when compared with males who were not faecally incontinent, see table 21.

Table 21– Sexual Satisfaction Scores in Sub-groups.

Using the sexual satisfaction subscale of the Brief Inventory of Sexual Function, those who are incontinence of urine and faeces were compared with those who were continence to see if there were any significant difference.

	Numbers	Mean Satisfaction Score	SexSD	p
Faecal Incontinence	9	2.3	1.62	0.44
Faecal Continence	19	2.74	1.28	
Urinary Incontinence	8	2.00	1.51	0.19
Urinary Continence	23	2.72	1.23	
Genital Anomalies	10	2.30	1.45	0.49
No Anomaly	21	2.65	1.23	

However in our group of male subjects only scores for the ‘Sexual Anxiety’ subscale of the MSQ were significantly different in males who were faecally incontinent compared with those with good or perfect continence ($p=0.03$).

Discussion

It is uncommon for doctors and parents to consider the consequences of genital anomalies when the affected individual is still a child. However, it is necessary to consider these issues prior to adolescence as not only should reconstructive surgery be carefully considered but the expectations of what is achievable should also be closely managed.

Genital anomalies

Associated anomalies with ARM are well documented. However, authors writing on surgical reconstruction in childhood may not be too concerned with the genitalia and may simply fail to record anomalies that are not directly relevant to their management (189). Hall et al, in trying to quantify the incidence of genital anomalies from a retrospective review, found that the vagina had been assessed in only 72 of 162 girls and the internal genitalia in only 51. They found that 22 of 72 (32%) had a vaginal anomaly and 18 of 51 (35%) had a uterine anomaly. All of the uterine anomalies were serious (47). Subsequent studies have found the rate of female genital anomalies to be lower (190), this may be in part explained by the delayed diagnosis of female genital anomalies as many of these do not present until puberty.

In the case of females with a cloaca where the levels of genital anomalies is higher, it is recommended that there is an assessment of the genital tract in puberty to uncover any anomalies that may have been missed in earlier assessments (191). Earlier reports have advised the resection of atretic Mullerian structures in females with cloaca (192), however it is possible these small and underdeveloped structures may develop during puberty. These patients should undergo an examination of the external genitalia, an internal examination should also be offered in those who have been sexually active. In addition a pelvic MRI is essential to assess any anomalies of the Mullerian structures.

In this study it was demonstrated that all males with associated genital anomalies and 12/14 females with genital anomalies had other anomalies. Children with a diagnosis of ARM with associated anomalies should be closely assessed for the presence of a genital anomaly either in childhood or in puberty in females. The consequences of a Mullerian anomaly are best addressed prior to attempts to become pregnant as the issue of increased rates of miscarriage can be discussed and prepared for.

An exact level of male genital anomalies is difficult to define because of limited and contradictory literature. Hoekstra et al recorded all of the genito-urinary anomalies associated with ARM in 150 children. The genital anomalies in boys were relatively uncommon and mainly of little significance, especially when considering sexual

function in later life (193). Cho et al recorded 16 anomalies in 69 boys (23%), assuming all of those with ambiguous genitalia were male (194). It would seem, therefore, that there is a wide range of possible genital anomalies in boys, many of which would have little impact on future sexuality or fertility.

Testicular maldescent is reported to be present in 3% to 19% of males with ARM (195;196), furthermore the higher the level of anorectal malformation the more likely the higher the rates of cryptorchidism. Cortes et al found that rates of cryptorchidism approached 50% in boys with associated renal or ureteric anomalies (197). However the rate of infertility attributable to undescended testes cannot be accurately determined and paternity rates reported in the literature for undescended testes ranges greatly (198). The consequences of maldescent of the testes include impaired fertility, this was also seen in at least one participant as a consequence of recurrent epididymitis, this has been previously reported (199).

Sexual Satisfaction

The difference that exists between male and female participants with regards to sexual satisfaction scores and sexual anxiety concurs with the authors own anecdotal experience. The reason for this is not clear. It may be postulated females are more worried about the impact of their ARM has on their partner in terms of attractiveness and sexual abilities. Whether this is secondary to body image or worries about incontinence it is not clear and may very well represent a combination of factors. The concept of urinary incontinence and sexual dysfunction is well documented in the literature (200). This demonstrated all areas of sexual functioning to be impaired in females with LUTS and UI, including hypoactive sexual desire. One of the limitations of comparing our group with those included in studies of this type are the age differences, as many of those included are post-menopausal and there may well be other factors contributing to their sexual dysfunction.

Spinal cord anomalies may have a major impact on sexual function in both genders, it tends to be more severe in males, particularly in those born with myelodysplasia. The

reconstructive surgery probably has little effect. The bladder problem is usually an upper motor neurone hyper-reflexia due to lumbosacral anomalies. In a review of the literature 29% of those with a high anorectal anomaly and 6% with a low anomaly had urinary incontinence (201). If an analogy can be drawn with the myelodysplasia population, the impact on potency may not be great especially if the neurological lesion is incomplete. All males with myelodysplasia with intact sacral reflexes and urinary continence are potent. With absent sacral reflexes, 64% with levels below D10 and 14% with levels above D10 are potent (202). As most of those with anorectal agenesis and neuropathy have a level well below D10, few should be impotent.

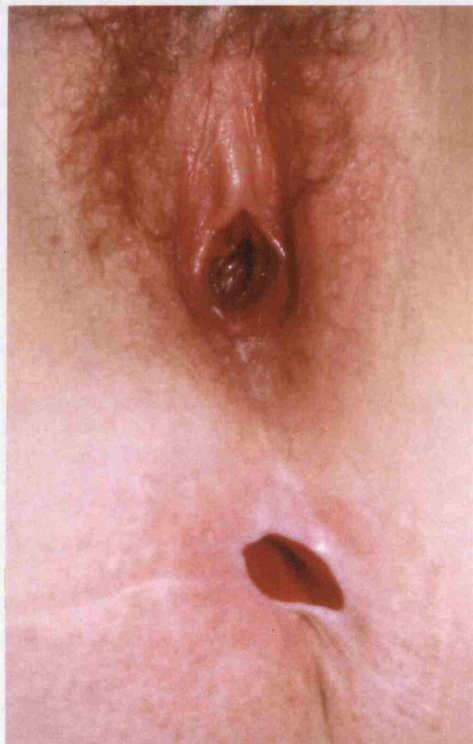
One study that looks more closely at this group by Holt et al found that in 20 patients investigated for infertility who had undergone an anorectal pull through procedure that iatrogenic injury accounted for at least half of the cases (203). Other reported causes included recurrent epididymo-orchitis (204), which may result as a consequence of urine refluxing into the vas deferens (205). This urinary reflux may be idiopathic or may be as a consequence of an associated vasal anomaly, such as vasal ectopia to bladder, large prostatic utricle, posterior urethral valves and complete duplication of ureteral system (206). Iatrogenic injury may also be implicated in urinary reflux. Epididymitis should be considered when ARM patients present with an acute scrotum as it may prevent unnecessary surgical exploration.

Rintala et al have compared their own patients with low and high anomalies separately and compared them to healthy controls. In those with a low anomaly 11 of 83 patients (13%) had difficulties with intercourse but always related to concerns about anal function. No mention was made of any patient being physically unable to have intercourse (45). In patients with an intermediate or high anomaly, ten of 33 (30%) had sexual dysfunction. Although it is not stated how many of these were male, three had erectile dysfunction and two had retrograde ejaculation. Six of the ten cited fear of anal incontinence as a significant limitation on their sexual activity (46). This is higher than the rates of increased sexual anxiety seen in our male participants with a degree of faecal incontinence.

The timing of vaginal reconstruction is important. If surgery is carried out in young girls, dilation may be required throughout childhood for an organ that will not be used for many years (see figure 15). Perhaps the most compelling argument against surgery in infancy is the risk of neoplasia. In a review, Schober identified five cases of squamous cell carcinoma of skin vaginas and four cases of adenocarcinoma of intestinal vaginas between 1927 and 1994. The cases occurred in women between 25 and 30 years old and between eight and 25 years after reconstruction (207). Average time from surgery to diagnosis is estimated to be 17 years (208). They are admittedly very rare cancers, nonetheless, the risk remains and a good case can be made for deferring elective surgery until the patient is competent to give her own consent.

Figure 15 – Photograph of a patient with a cloaca anomaly.

She has an end ileostomy and is using her redundant rectal pull-through as a vagina. The appearance is unsatisfactory as her perineum is scarred from previous surgery, and there are no labia or introitus.



Conclusions

Anorectal malformations have adverse implications for fertility and sexual function in both men and women. Full information about the diagnosis and its implications must be made available to the family and should be explained clearly so that the patients, if old enough and their parents can fully appreciate what the problem is and what future holds for them. When dealing with adolescents and young adults with ARM it is important that issues regarding sexual function and fulfilment are addressed openly and without embarrassment. Many of these patients will have become used to visiting their paediatric surgeon or urologist where much of these adult problems are not fully addressed. It is important that these patients feel able to request help with issues of a more personal nature.

The majority of our participants are sexually active, and the mean of sexual debut does not appear to be that different from the normal population which is reported to be 17 years for both males and females (209). Furthermore, in those that were not sexually active corrective vaginal surgery was required to allow penetrative sexual intercourse. It is important for females with an ARM to be routinely assessed in adolescence by a gynaecologist to ensure that these are addressed without delay. This may reduce the amount of anxiety that these young people have regarding intimate relationships.

Chapter 8- Psychological Outcomes

Introduction

The diagnosis of a serious medical condition at birth and subsequent repeat protracted hospital admissions can be expected to have psychological consequences on some patients. Absences from school, demanding treatment regimens, parental anxiety, invasion of personal privacy, pain or physical discomfort, can all impact upon socialization and family relationships.

In general, increased levels of psychological difficulties have been identified for a range of diagnoses of chronic illness, including a reduction in quality of life (210), so that it has been recommended that clinical management of chronic illness should extend to psychosocial difficulties. There is evidence to suggest that parents relate to children with chronic illness differently compared to siblings. Where this persists into adolescence, an individual may be at risk of remaining unduly dependent upon their parents (211) so that transition to adulthood could pose another type of challenges for patients and parents. In a recent study that addressed the specific concerns of parents of ARM children, 40% reported worries about the implications of the child's ability to form and maintain relationships in adulthood, engage in sexual intimacy and find gainful employment (212).

In another study involving 33 adolescent patients (213), significant levels of psychological disturbance was identified in 58% of the sample, higher than the 35% reported in an earlier study (63). Greater levels of disturbance in the former were attributed to the twice daily anal dilatations that the patients had had to undergo as children. The intrusiveness of repeat anal dilatation by adults may distort adult-child relationships and well being. It may also compromise the development of a positive body image and sexual confidence. Interpretation of any psychological data should thus take account of bowel management regimes for young patients with ARM, where rectal enema administration may be required to achieve faecal continence.

Whilst the hypothesised relationship between increased psychological disturbance and incontinence has some credence, it is not always supported in reports. For example some studies have suggested that levels of faecal incontinence may not be linearly related to levels of psychological maladjustment (63;214). This contention was contradicted by a subsequent report (215), in which 66.7% of children with poor bowel control had behavioural problems compared with only 8.6% in those considered to have good bowel control. It was further suggested that the psychological sequelae of faecal incontinence would have a lasting effect into adult life, influencing key aspects of life including occupation and relationships.

Overall, very little data exist on psychological outcomes in adults with the diagnosis of ARM other than a limited literature on sexual relationships and satisfaction (216). Furthermore the focus has generally been on the paediatric/adolescent age groups. The results of the current study contribute to further knowledge.

Patients and Methods

As stated in the Methods chapter the participants were required to complete questionnaires in Quality of Life and Body Esteem. Quality of life was assessed using the SF-36 questionnaire. This is a validated question and contains 36 items which measure eight dimensions; physical functioning, social functioning, role limitations due to physical problems, role limitation due to emotional problems, mental health, energy/vitality, pain and general health perception. Body Esteem was assessed utilising Body esteem refers to the overall positive or negative evaluation of the body by and individual (137). Poor body esteem is felt to be prevalent in people with disabilities and chronic disease states. Furthermore this may be compounded by the fact that society as a whole perceives the bodies of the disabled in a negative way (138).

As many of the scoring systems were gender specific it was deemed necessary to find a tool that could be used in both males and females, to allow a comparison to be made. The only tool that seemed suitable for this was the Body Esteem Scale (217;218). This scale was developed specifically for use on young adult populations, and allows a means of not only comparison within our sample population but also with published norms.

Results

Only 1 subject revealed that she had a formal diagnosis of depression, and required regular medication for this. On questioning, she did not relate her depression specifically to her diagnosis of ARM, which was a low anomaly with no associated congenital abnormalities. This patient was in full-time employment and lived with a partner. None of the other subjects reported having received formal help for psychological difficulties or mental health problems.

As reported in chapter 4, the majority of the subjects were in full-time employment or were students. Only 9 described themselves as being registered disabled, of these 2/9 had low ARM's, 3/9 had high anomalies, 3/9 (all females) had a diagnosis of cloaca, and for 1/9 there were no medical notes available to ascertain an accurate diagnosis. Again, of the 9 registered disabled subjects, 4/9 also had associated anomalies of the VACTREL association. All 9 were, or had been, sexually active and 4 of them were married or living with partner.

Body Esteem

There is little data on body image in patients with a diagnosis of ARM. The scoring system utilised in this study is the Franzoi and Shields (219). The score has three subscales for males: Physical Condition (PC), Physical Attractiveness (PA) and Upper Body Strength (UBS). There are three similar subscales for females which are Physical Condition (PC), Sexual Attractiveness (SA) and Weight Control (WC). The scoring system gives higher scores to those with better outcomes. There are normal values for each of these subscales in the literature and our results were compared to these.

The results for males in 2 of the 3 subscales; Upper Body Strength and Physical Condition, the results scored by individuals with ARM were significantly lower than those scored by the normal controls. The only subscale where the results were not significantly different was Physical Attractiveness (see table 22).

Table 22 – Body Esteem Results for Males

Male participants were compared to published normal data. The scores for the ARM group were lower across all 3 sub-scales.

	ARM n=31		Normal controls n=331		p=
	Mean	SD	Mean	SD	
Physical Attractiveness	37.13	7.10	39.1	5.7	0.07
Upper Body Strength	29.03	5.80	34.0	6.1	0.0001
Physical Condition	41.52	9.16	50.2	7.7	0.0001

The female participants in the current study yielded significantly lower scores compared to normal values across all three subscales, suggesting that females with a diagnosis of ARM are more likely to report negative body esteem compared to females in the general population (see table 23). There are many factors that may contribute to body esteem, and the results were further analysed to assess if there were factors other than the diagnosis of ARM that affected the outcomes. Those patients from socioeconomic classes I and II had higher scores which are suggestive of a lesser degree of impairment for Weight Concern and Physical Condition compared to participants from socio-economic classes III and lower. This difference was found to be significant using an unpaired student t-test ($p=0.02$ and $p=0.007$ respectively). Interestingly when the Physical/Sexual Attractiveness subscale was analysed, those in socioeconomic classes III and above had a mean score of 41.41 compared to those in classes I and II whose mean score was 39.16, though this difference was not significant, ($p=0.24$).

Table 23– Body Esteem Results for Females

Female participants were compared to published normal data. The scores for the ARM group were lower across all 3 sub-scales.

	ARM n=44		Normal controls n=633		p=
	Mean	SD	Mean	SD	
Sexual Attractiveness	43.35	8.49	46.9	6.3	0.0005
Weight Concern	27.25	8.42	29.9	8.2	0.0389
Physical Condition	27.93	7.75	33.3	5.7	0.0001

The Body Esteem data were further examined to see if there was a correlation between the underlying level of the malformation and body esteem. Interestingly there were no significant differences in the subscale scores between those with low ARM and high ARM. The only significant finding was in the Physical Conditioning subscale when those with a diagnosis of high ARM and cloaca were compared. Possible reasons for this include the fact the cloaca group is entirely female, and females have a more negative perception of their body image or that the numbers involved are very small. When only females with a diagnosis of high ARM (n = 8) and cloacas were compared no significant difference was found.

Quality of Life

As discussed in Chapter 3 quality of life was assessed using the Short Form 36 questionnaire comprising 8 subscales measuring: Physical Functioning, Social Functioning, Role Limitation due to Physical Problems, Role Limitation due to Emotional Problems, Mental Health, Energy/Vitality, Pain and General Health Perception. These are all aspects of quality of life which one could anticipate being impaired by a chronic illness. Item scores for each item are coded and transformed to

give an overall score from 0 (worst possible) to 100 (best possible). Also included in the questionnaire is an additional question which assesses any recent change to health.

These results were compared with previously published normal values which are derived from American populations. The results of the participants differed significantly only for the subscales General Health and Role Limitation-Emotional, $p < 0.0001$, and $p = 0.02$ (see table 24).

Table 24 - Summary of SF-36 results

Scores for all eight dimensions of the SF-36 for the ARM group are compared with normal control data. In only 2 sub-scales; General Health and Role limitation – social are there significant differences.

	ARM Mean n=75	ARM SD	Normals Mean n=2479	Normals SD	p=
Physical Functioning	84.40	23.29	84.5	22.9	0.97
Role limitation – physical	84.80	33.32	81.2	33.8	0.08
Bodily Pain	76.72	27.35	75.5	23.6	0.66
General Health	57.55	26.78	72.2	20.2	<0.0001
Vitality	62.07	20.31	61.1	20.9	0.69
Social Functioning	84.34	20.96	83.6	22.4	0.78
Role limitation – Emotional	74.32	38.44	81.3	33.0	0.02
Mental Health	71.88	17.75	74.8	18.0	0.17

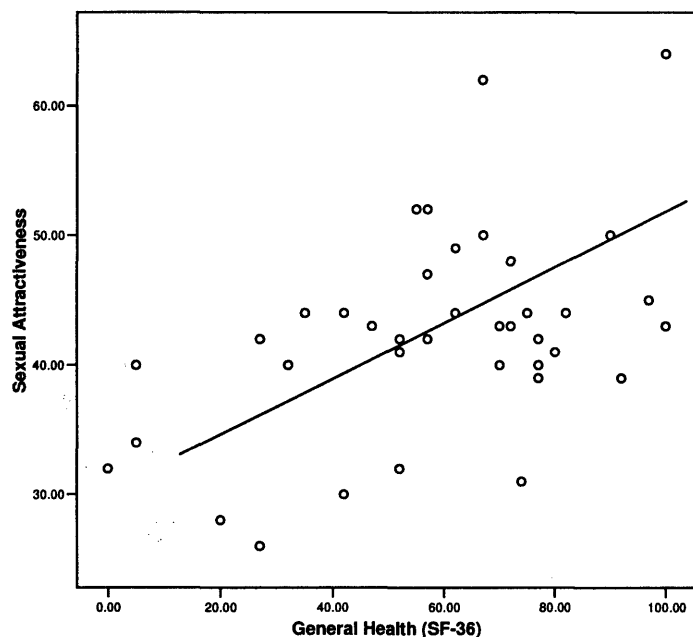
Further analysis of the SF-36 subscales has yielded a positive correlation between Bodily Pain, General Health and Social Functioning when correlated against Physical Attractiveness using the Pearson Rank test. There was a moderate positive correlation was between Bodily Pain and Sexual Attractiveness ($r=0.303$, $p=0.009$). However, when the General Health scores were correlated against Sexual Esteem and Sexual

Satisfaction no significant association was demonstrated ($p=0.33$ and 0.56 respectively). Thus individuals may feel less sexually attractive but this may not necessarily impair sexual satisfaction.

Sexual attractiveness as measured by the Body Esteem Score and General Health as measured by the SF-36 were correlated to see if one could predict those individuals who generally felt healthier also felt more sexually attractive. A scatter plot shown below (figure 16) demonstrates a positive correlation between the two.

Figure 16 – Scatter Plot of Sexual Attractiveness and General Health

Scatter plot and trend line demonstrating the improving general health for males and females with ARM has a positive effect on sexual attractiveness.



These SF-36 subscale scores were further analysed against the level of malformation. Interestingly, there were no significant difference between those with a diagnosis of low ARM and those with high ARM on all subscales. The only result which neared significance was on the subscale looking at Mental Health, ($p=0.057$).

Impact of condition on ability to form relationships

The results were analysed to see if there was a relationship between incontinence, the method of micturition or the presence of a stoma and relationship status. Using a 2-tailed student t-test, no significant relationship has been found between those that were in a relationship and those that were not ($p=0.53$). A similar result was found when the method of micturition was looked at, those that either performed CISC or had some form of urinary diversion were as likely to be in a relationship as those that voided normally ($p=0.548$). Furthermore, the presence of a bowel stoma did not appear to have affect relationship status. Of those individuals with a stoma 28% were married, compared with only 16% if those without a stoma.

One variable that appeared to have some impact on relationship status was socioeconomic class, with those in SEC 1 or 2 significantly more likely to be in a relationship than those in SEC 3-5 ($p=0.004$). The reason for this was not clear, as other factors such as age, gender, and underlying level of malformation did not appear to be different between these two groups.

Discussion

Overall, given the considerable physical and psychological challenges of living with ARM, participants of the current study have reported few major problems in Body Esteem and Quality of Life. By contrast, significant psychological maladjustment has been reported for paediatric populations (63;220). One possible reason for this discrepancy is that the school environment can be intimidating for children with disabilities including incontinence and that coping mechanisms have come into play by adulthood.

Methodological differences between studies can also influence results. Several studies involved interviewing parents and carers as well as the affected individual. The current study has relied upon self completed questionnaires. This method has been reported to be more accurate in assessing potentially embarrassing symptoms (142). With hindsight, it would have been useful to have included an assessment of global psychological well being. Since not everyone experiencing psychological distress

would seek formal help, such an assessment would have given an indication of psychological well being.

Concerns regarding the lack of formal psychological support offered to patients with ARM and parents have been highlighted in a recent questionnaire based study (221). From the authors own experience with these individuals, there is very little in terms of routine psychological support offered to adults with ARM. This is in part due to the relative lack of availability of such services, and suitably qualified staff to offer the specialist interventions where needs be. However, it is important to highlight that of those that attended for interview none requested specific psychological input, and none voiced concern about neglect over this aspect of their care.

Body Esteem

Body esteem is an important part of self-esteem and is often an overlooked aspect in long-term outcome data, perhaps because it can be difficult to quantify. However, in our increasingly appearance conscious society, it is important to appreciate how experiences of ARM might impact upon body esteem.

Whilst the score utilised in this study had its limitations it does at least represent an attempt to quantify various aspects of an individual's body esteem. A further limitation of the scoring system utilised is that it did not allow direct comparison between the male and female subjects, which may have proved insightful.

The reason for higher scores in the Physical Condition, Weight Concern and Upper Body Strength subscales in those of SEC I and II is unclear. There are no normal values with demographic data to compare these results with. In general, populations with higher socio-economic status enjoy greater levels of psychological well being and health related quality of life. This also extends to parental socioeconomic class of children and adolescents (222). Other factors such as parental educational levels are also reported to influence psychological wellbeing. Unfortunately this was not assessed in this survey but may have provided some useful additional information.

However there is also evidence to suggest that females in higher socioeconomic classes are more likely to express dissatisfaction with image, in particular weight

(223). Therefore presumably the relationship between SEC and body image is complicated. There are data to suggest that ethnicity also has a role to play in perceived ideal weight, with white females being more likely to perceive themselves as being overweight (224). The fact that those in SEC III and above had higher scores for the Sexual/Physical Attractiveness subscales suggests that the relationship between SEC and body esteem is more complicated than simply increased SEC equals increased self esteem. Other compounding factors not considered as part of this study must also have a role to play in this complex relationship.

Quality of Life

The possibility of chronic disease having a negative effect upon quality of life has been well documented. However when compared to the reported “normal” population, our subjects only differed significantly on the subscales looking at General Health and Role Limitation – Emotional. Whether this is a true difference or an artefact of demographic differences is not clear. An improvement in the study design would have involved enrolling age matched normal controls from the United Kingdom. It is not difficult to see the why some of the affected individuals have specific problems with emotional aspects of their quality of life. Whether this limitation is due to the consequences of ARM, such as incontinence, rather than the diseases itself is also a possibility (63).

Other studies looking specifically at quality of life have found that their ARM population had good overall scores when compared to a normal population (225). In our subjects it was demonstrated that those with poor perceived general health, as quantified by the SF-36 questionnaire, had decreased scores on the sexual attractiveness sub-scale. However no similar decrease in sexual esteem or satisfaction was observed. Therefore it is possible that improving overall perception of health, but perhaps not necessarily any specific health parameters may have a positive impact on other aspects of these individuals functioning.

Impact of condition on ability to form relationships.

Previously published data has reported that these individuals have difficulty forming close personal relationships. Hassink et al reported that almost a quarter of adult individuals with a high ARM had never had a relationship, with the majority of these ascribing this to their medical diagnosis (225). The data presented in this study is far more optimistic with the majority of individuals in relationships and sexually active.

Possible reason for the differences observed include the fact that only individuals with a high ARM were looked at. However if we consider only those with a documented high ARM then 20/26 (77%) have been sexually active, and 16/26 (62%) are currently in a relationship. Of the 6 that were not sexually active in our study; one female was referred for surgery to treat vaginal stenosis, and one male had significant rectal prolapse and was awaiting surgery to reduce this. Two further individuals had significant associated anomalies; VATER and spina bifida, which would undoubtedly have a negative affect on an individual's ability to form and maintain a personal relationship.

Conclusions

ARM does not appear to be associated with significant problems of Body Esteem and Quality of Life. Parents may be reassured that a child born with the diagnosis of ARM is capable of living independently, without major psychological difficulties. Despite less than perfect continence outcomes, affected individuals are capable of a reasonable quality of life and psychological well being.

Any psychological intervention deemed necessary could strategically target issues shown to be of concern, such as Body Esteem in adult women with ARM. In our appearance conscious culture the "lucky to be alive" attitude held by many of these individuals and their families will increasingly diminish as morbidity and mortality results for babies born with an ARM improve. Therefore it will become increasingly important to address issues regarding body image and quality of life.

Chapter 9 – Conclusions

The overwhelming impression of this study is one of optimism. The majority of individuals that took part were genuinely optimistic about their situation and what the future held for them. Most were interested in their condition and almost all requested information about the treatment that they had received as children. However, the overall clinical outcomes of those born with an anorectal malformation are in many respects disappointing. A significant number of individuals were incontinent of urine and of faeces. Nevertheless, the majority even of those with relatively poor outcomes in terms of urinary and faecal continence were in relationships, employment and are sexually active.

Bowel Function

The majority (59%) of participants were able to defecate per rectum without any form of assistance. Eighteen (24%) had some form of stoma, the majority of whom had an ACE. Seven participants (9%) had permanent stomas, either ileostomy or colostomy. Overall, these results for faecal incontinence are comparable to those published in the wider medical literature. Incontinence was assessed using the Cleveland Clinic Incontinence Score. Fifty seven percent of those assessed reported themselves as having perfect or good bowel control. Twenty one (31%) of the participants described themselves as having moderate incontinence and 8 (12%) reported having severe incontinence.

Predictably those individuals with more severe malformations tended to fair worse in terms of bowel control (see Table 9). Furthermore, there did not seem to be an appreciable difference in the incontinence rates when those who had undergone a PSARP repair were compared with those individuals who had a pullthrough procedure. However, only 7/75 had undergone a PSARP as their initial repair. With improved surgical techniques it is probable that follow-up studies of children who have undergone the PSARP procedure as adults will have different outcomes than those reported in this study.

Other predictors for incontinence included having undergone an ACE procedure. Of the 11 individuals with an active ACE stoma only 2 (18%) described themselves as having good continence. In addition those individuals with an ACE scored worse in terms of quality of life when compared to individuals with permanent stomas or those practising regular rectal washouts (see table 11).

Surprisingly only 3 participants described themselves as constipated (as assessed by the Cleveland Clinic Constipation Score). This is in contrast to previously published data in the literature (226). There was no simple explanation for this surprising outcome.

Urinary Function

Renal anomalies were detected in 53% of participants, the majority of whom had vesicoureteric reflux. This is comparable to other studies. The majority of patients void spontaneously and 60% are dry. Eight participants had some form of urinary diversion, 7 had Mitrofanoffs and one had an ileal conduit. Unfortunately 6/7 (86%) of those with a Mitrofanoff were incontinent of urine, most on a daily basis. Again the rates of incontinence seen were higher in those with the more severe malformations. The majority (73%) of those with a diagnosis of cloaca had significant incontinence. 15% of those with a diagnosis of high ARM were incontinent on a daily basis. Perhaps more surprisingly was the finding of incontinence on a daily or weekly basis in those with a diagnosis of low ARM. Previous reports in the literature have reported total continence in this group of individuals (227).

Sixteen of the 75 (21%) reported urinary frequency, the majority of who were females (12/16). The majority of those affected did not find this to be a particularly bothersome symptom. In addition nocturia was found in 10/75 (13%), all of whom found this to be bothersome.

There is evidence to suggest that those individuals with an abnormal bladder in childhood who undergo surgery can expect a deterioration in adolescence (228) and it may be that when other centres look specifically at their adult outcome data that a similar picture may emerge. In addition, this study involved clinicians not directly

involved in the participant's current or previous medical care, which may have eliminated any reporting bias and gives a more objective view of long-term outcomes.

It is possible that improved modern surgical techniques minimise the risk of urinary incontinence. However, this is difficult to confirm as no studies have directly compared adult patients who have undergone one type of repair with the PSARP.

Sexual Function & Fertility

The results regarding sexual satisfaction and body image demonstrate that despite considerable morbidity associated with diagnosis, treatments and associated anomalies, the majority of our participants are functioning well both physically and psychologically. As this is the largest series of ARM individuals in the UK, where this aspect of functioning has been fully assessed this aspect should be highlighted to parents with affected children. When dealing with adolescents and young adults with ARM it is important that issues regarding sexual function and fulfilment are addressed openly and without embarrassment. Many of these patients will have become used to visiting their paediatric surgeon or urologist where much of these adult problems are not fully addressed.

Eighty seven percent of our participants are sexually active, and the mean of sexual debut does not appear to be that different from the normal population (209). Two female participants, required corrective vaginal surgery to allow penetrative sexual intercourse. These individuals had been lost to specialist follow-up although why this occurred was unclear. The consequences of a Mullerian anomaly in a female are best addressed prior to attempts to become pregnant as the issue of increased rates of miscarriage can be discussed and prepared for. Therefore it is recommended that females diagnosed with ARM should be routinely assessed in adolescence by a gynaecologist. This is to ensure that any problems are addressed without delay. This may also help reduce the amount of anxiety that these young people have regarding intimate relationships.

Despite the frequency of associated genital anomalies, fertility outcomes were encouraging in the male patients. 11 of the 31 men (35%) had partners who had

conceived. On initial inspection of our data, fertility rates for women appear lower with only 7 of the 44 women (16%) conceiving. However this group of individuals are young, mean age 25 years and the mean age for first pregnancy in the UK population is currently 29 years (229). Only 8 of the women had attempted conception giving a pregnancy rate of 88%. It is very encouraging that all but three of the women menstruate normally and the majority were sexually active. The high number of genital anomalies may contribute to infertility, miscarriage and preterm delivery and further follow-up of this cohort of women should provide more accurate pregnancy data.

Psychological Function

The results of this study demonstrate that the ARM and cloaca population do not have high levels of psychological problems and mental health issues despite a childhood of repeated surgical interventions. Therefore parents should be reassured that their children are likely to have near normal adult lives, with the expectation that their children should be able to develop into independent individuals. Despite less than perfect continence outcomes, affected individuals are capable of a reasonable quality of life and psychological well being.

Only one individual described themselves as depressed and was taking medication for this. None of the other participants were having any treatment for mental health problems. This is surprising as one would anticipate a greater number of affected individuals even amongst the “normal” population mental health problems exist in approximately 1 in 6 (230). Possible explanations for this include the fact that those with a diagnosis of ARM are psychologically well adjusted, and less susceptible to mental health problems.

Any psychological intervention deemed necessary could strategically target issues shown to be of concern, such as Body Esteem in adult women with ARM. In our appearance conscious culture the “lucky to be alive” attitude held by many of these individuals and their families will increasingly diminish as morbidity and mortality results for babies born with an ARM improve. Therefore it will become increasingly important to address issues regarding body image and quality of life.

Recommendations for future work

In order to maintain a complete data set on these patients it would be beneficial to set up a nationwide database of all babies born with a diagnosis of anorectal malformation. Ideally this would be coordinated by a central organisation, such as the British Association of Paediatric Surgeons. This would allow prospective data collection with more effort to unify diagnosis criteria, investigations and management protocols. This would allow analysis of long-term outcomes to be performed at regular intervals, and direct comparisons with outcomes in other countries. Furthermore the outcomes of specific operative procedures and treatments could be more easily compared.

Setting up of such a database would be no small undertaking and would require the cooperation of all paediatric surgeons and healthcare professionals involved in the management of these patients. However, experience with cancer care has demonstrated that it is possible to achieve such a database with a few individuals in each centre responsible for data entry, and a single body or centre to oversee the running of the database. This will allow outcomes to be accurately reported without some of the inevitable biases and difficulties encountered by the researchers of this study

In addition to setting up a central database, there is also a need for individual centres and indeed individual surgeons to maintain accurate information on their patients with regards to the precise nature of the anomaly, any associated anomalies and treatment. In an increasingly litigious society it is imperative that surgeons take measures to ensure that medical records are accurate and secure.

Other helpful resources that may be of particular benefit to these patients include patient support groups. These can provide useful additional information, often of a more practical nature, details of such organisations should be available in the clinics where these patients are seen. However the emphasis put on parent and patient support groups by paediatric surgeons is in some cases limited (231). This it is

postulated is partly due to lack of appreciation of the positive impact that some of these groups have.

Bibliography

- (1) Pena A. Anorectal malformations. *Semin Pediatr Surg* 1995 Feb;4(1):35-47.
- (2) Office for National Statistics. Live Births in England and Wales, 2004: area of residence, population trends. 2004.
- (3) Hendren WH. Repair of cloacal anomalies: current techniques. *Journal of Pediatric Surgery* 1986;21:1159-76.
- (4) Taipale P, Rovamo L, Hiilesmaa V. First-trimester diagnosis of imperforate anus. *Ultrasound Obstet Gynecol* 2005 Feb;25(2):187-8.
- (5) Veyrac C, Couture A, Saguintaah M, Baud C. MRI of fetal GI tract abnormalities. *Abdom Imaging* 2004 Jul;29(4):411-20.
- (6) Lam YH, Shek T, Tang MH. Sonographic features of anal atresia at 12 weeks. *Ultrasound Obstet Gynecol* 2002 May;19(5):523-4.
- (7) Harris RD, Nyberg DA, Mack LA, Weinberger E. Anorectal atresia: prenatal sonographic diagnosis. *AJR Am J Roentgenol* 1987 Aug;149(2):395-400.
- (8) Warne S, Chitty LS, Wilcox DT. Prenatal diagnosis of cloacal anomalies. *BJU Int* 2002 Jan;89(1):78-81.
- (9) Moore KL, Persaud TVN. *Before We Are Born: Essentials of Embryology and Birth Defects*. 6 ed. Saunders; 2003.
- (10) Penington EC, Hutson JM. The absence of lateral fusion in cloacal partition. *J Pediatr Surg* 2003 Sep;38(9):1287-95.
- (11) Thomas DFM. *Cloacal Malformations: Embryology, Anatomy and Principles of Management*. Berlin & Heidelberg: Springer-Verlag; 1989.
- (12) Nievelstein RA, van der Werff JF, Verbeek FJ, Valk J, Vermeij-Keers C. Normal and abnormal embryonic development of the anorectum in human embryos. *Teratology* 1998 Feb;57(2):70-8.
- (13) Cuschieri A, EUROCAT Working Group. Anorectal Anomalies Associated With or as Part of Other Anomalies. *American Journal of Medical Genetics* 2002 Feb;110:122-30.
- (14) Czeizel A, Ludanyi I. An aetiological study of the VACTERL-association. *Eur J Pediatr* 1985 Nov;144(4):331-7.

- (15) Falcone RA, Jr., Levitt MA, Pena A, Bates M. Increased heritability of certain types of anorectal malformations. *J Pediatr Surg* 2007 Jan;42(1):124-7.
- (16) Myers MF, Li S, Correa-Villasenor A, Li Z, Moore CA, Hong SX, et al. Folic Acid Supplementation and Risk for Imperforate Anus in China. *Am J Epidemiol* 2001 Dec 1;154(11):1051-6.
- (17) Cuschieri A, EUROCAT Working Group. Anorectal Anomalies Associated With or as Part of Other Anomalies. *American Journal of Medical Genetics* 2002 Feb;110:122-30.
- (18) Martinez-Frias ML, Frias JL, Opitz JM. Errors of morphogenesis and developmental field theory. *Am J Med Genet* 1998 Apr 1;76(4):291-6.
- (19) Stephens FD. Wingspread anomalies, rarities, and super rarities of the anorectum and cloaca. *Birth Defects Orig Artic Ser* 1988;24(4):581-5.
- (20) Pena A. Anorectal malformations. *Semin Pediatr Surg* 1995 Feb;4(1):35-47.
- (21) Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW, III. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. *J Urol* 1997 Sep;158(3 Pt 2):1298-300.
- (22) Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW, III. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. *J Urol* 1997 Sep;158(3 Pt 2):1298-300.
- (23) McLorie GA, Sheldon CA, Fleisher M, Churchill BM. The genitourinary system in patients with imperforate anus. *J Pediatr Surg* 1987 Dec;22(12):1100-4.
- (24) Boemers TM, de Jong TP, van Gool JD, Bax KM. Urologic problems in anorectal malformations. Part 2: functional urologic sequelae. *J Pediatr Surg* 1996 May;31(5):634-7.
- (25) Warne SA, Wilcox DT, Ledermann SE, Ransley PG. Renal outcome in patients with cloaca. *J Urol* 2002 Jun;167(6):2548-51.
- (26) Misra D, Mushtaq I, Drake DP, Kiely EM, Spitz L. Associated urologic anomalies in low imperforate anus are capable of causing significant morbidity: a 15-year experience. *Urology* 1996 Aug;48(2):281-3.
- (27) Hong AR, Acuna MF, Pena A, Chaves L, Rodriguez G. Urologic injuries associated with repair of anorectal malformations in male patients. *J Pediatr Surg* 2002 Mar;37(3):339-44.
- (28) Rintala RJ, Lindahl HG. Fecal continence in patients having undergone posterior sagittal anorectoplasty procedure for a high anorectal malformation improves at adolescence, as constipation disappears. *J Pediatr Surg* 2001 Aug;36(8):1218-21.
- (29) Pena A. Anorectal malformations. *Semin Pediatr Surg* 1995 Feb;4(1):35-47.

- (30) Hibi M, Iwai N, Kimura O, Sasaki Y, Tsuda T. Results of Biofeedback Therapy for Fecal Incontinence in Children with Encopresis and Following Surgery for Anorectal Malformations. *Dis Colon Rectum* 2003 Oct;46(10(suppl)):54-8.
- (31) Pena A. Anorectal Malformations: Experience with the Posterior Sagittal Approach. In: Mark D, Oldham K T, Mouriquand P, Edward R, editors. *Pediatric Surgery and Urology: Long Term Outcomes*. 1 ed. Saunders; 1998. p. 376-86.
- (32) Pena A, Levitt M, Hong AR, Midulla P. Surgical Management of Cloacal Malformations: A Review of 339 Patients. *J Pediatr Surg* 2004 Mar;39(3):470-9.
- (33) Hassink EA, Rieu PN, Brugman AT, Festen C. Quality of life after operatively corrected high anorectal malformation: a long-term follow-up study of patients aged 18 years and older. *J Pediatr Surg* 1994 Jun;29(6):773-6.
- (34) Davies MC, Creighton SM, Wilcox DT. Long-term outcomes of anorectal malformations. *Pediatr Surg Int* 2004 Aug;20(8):567-72.
- (35) Iwai N, Yanagihara J, Tokiwa K, Deguchi E, Takahashi T. Results of surgical correction of anorectal malformations. A 10-30 year follow-up. *Annals of Surgery* 1998 Feb;207(2):219-22.
- (36) Ackroyd R, Nour S. Long-term faecal continence in infants born with anorectal malformations. *J R Soc Med* 1994 Nov;87(11):695-6.
- (37) Iwai N, Hashimoto K, Goto Y, Majima S. Long-term results after surgical correction of anorectal malformations. *Z Kinderchir* 1984 Feb;39(1):35-9.
- (38) Ackroyd R, Nour S. Long-term faecal continence in infants born with anorectal malformations. *J R Soc Med* 1994 Nov;87(11):695-6.
- (39) Liem NT, Hau BD. Long-term follow-up results of the treatment of high and intermediate anorectal malformations using a modified technique of posterior sagittal anorectoplasty. *Eur J Pediatr Surg* 2001 Aug;11(4):242-5.
- (40) Tsuji H, Okada A, Nakai H, Azuma T, Yagi M, Kubota A. Follow-up studies of anorectal malformations after posterior sagittal anorectoplasty. *J Pediatr Surg* 2002 Nov;37(11):1529-33.
- (41) Ackroyd R, Nour S. Long-term faecal continence in infants born with anorectal malformations. *J R Soc Med* 1994 Nov;87(11):695-6.
- (42) Liem NT, Hau BD. Long-term follow-up results of the treatment of high and intermediate anorectal malformations using a modified technique of posterior sagittal anorectoplasty. *Eur J Pediatr Surg* 2001 Aug;11(4):242-5.
- (43) Hendren WH. Cloaca, the most severe degree of imperforate anus: experience with 195 cases. *Ann Surg* 1998 Sep;228(3):331-46.

- (44) Warne S, Wilcox DT, Creighton S, Ransley PG. Long-term gynecological outcome of patients with persistent cloaca. *Journal of Urology* 2003;170:1493-6.
- (45) Rintala R, Mildh L, Lindahl H. Fecal continence and quality of life in adult patients with an operated low anorectal malformation. *Journal of Pediatric Surgery* 1992;27:902-5.
- (46) Rintala R, Mildh L, Lindahl H. Fecal continence and quality of life in adult patients with an operated high or intermediate anorectal malformation. *Journal of Pediatric Surgery* 1994;29:777-80.
- (47) Hall R, Fleming S, Gysler M, McClorie GA. The genital tract in female children with imperforate anus. *American Journal of Obstetrics and Gynecology* 1985;151:169-71.
- (48) Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW, III. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. *J Urol* 1997 Sep;158(3 Pt 2):1298-300.
- (49) Laufer MR. Congenital absence of the vagina: in search of the perfect solution. When, and by what technique, should a vagina be created? *Curr Opin Obstet Gynecol* 2002 Oct;14(5):441-4.
- (50) Davies MC, Creighton SM, Woodhouse CR. The pitfalls of vaginal construction. *BJU Int* 2005 Jun;95(9):1293-8.
- (51) Greenberg JA, Wu JM, Rein MS, Hendren WH. Triplets after cloacal malformation repair. *J Pediatr Adolesc Gynecol* 2003 Feb;16(1):43-4.
- (52) Iwai N, Yanagihara J, Tokiwa K, Deguchi E, Takahashi T. Results of surgical correction of anorectal malformations. A 10-30 year follow-up. *Annals of Surgery* 1998 Feb;207(2):219-22.
- (53) Hendren WH. Cloaca, the most severe degree of imperforate anus: experience with 195 cases. *Ann Surg* 1998 Sep;228(3):331-46.
- (54) Greenwell TJ, Venn SN, Creighton S, Leaver RB, Woodhouse CR. Pregnancy after lower urinary tract reconstruction for congenital abnormalities. *BJU Int* 2003 Nov;92(7):773-7.
- (55) Landau D, Mordechi J, Karplus M, Carmi R. Inheritance of Familial Congenital Isolated Anorectal Malformations: Case report and review. *Am J Med Genet* 1997;71:280-2.
- (56) Boocock G R, Donnai D. Anorectal malformation: familial aspects and associated anomalies. *Archives of Disease in Children* 1987;62:576-9.
- (57) Cozzi F, Wilkinson AW. Familial incidence of congenital anorectal anomalies. *Surgery* 1968;64(3):669-71.

- (58) Stoll C, Alembik Y, Dott B, Roth MP. Evaluation of prenatal diagnosis of congenital gastro-intestinal atresias. *European Journal of Epidemiology* 1996;12:611-6.
- (59) Holt B, Pryor JP, Hendry WF. Male infertility after surgery for imperforate anus. *J Pediatr Surg* 1995 Dec;30(12):1677-9.
- (60) Holt B, Pryor JP, Hendry WF. Male infertility after surgery for imperforate anus. *J Pediatr Surg* 1995 Dec;30(12):1677-9.
- (61) Breslau N. Psychiatric disorder in children with physical disabilities. *J Am Acad Child Psychiatry* 1985 Jan;24(1):87-94.
- (62) Diseth TH, Emblem R. Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. *J Pediatr Surg* 1996 May;31(5):638-43.
- (63) Ludman L, Spitz L, Kiely EM. Social and emotional impact of faecal incontinence after surgery for anorectal abnormalities. *Archives of Disease in Children* 1994 Sep;71(3):194-200.
- (64) Bai Y, Yuan Z, Wang W, Zhao Y, Wang H, Wang W. Quality of life for children with fecal incontinence after surgically corrected anorectal malformation. *J Pediatr Surg* 2000 Mar;35(3):462-4.
- (65) Hartman E, Oort F, Visser M, Sprangers M, Hanneman M, Langen Z, et al. Explaining Change Over Time in Quality of Life of Adult Patients With Anorectal Malformations or Hirschsprung's Disease. *Diseases of the Colon & Rectum* 2006 Feb.
- (66) Dunn PM. Paulus Aegineta (625-690 AD) and Byzantine medicine. *Arch Dis Child Fetal Neonatal Ed* 1997 Sep 1;77(2):F155-F156.
- (67) Adams F. The seven books of Paulus Aegineta. Vols. I-III. 1844.
- (68) Wangenstein OH, Rice CO. Imperforate Anus: a method of determining the surgical approach. *Ann Surg* 1930;92:77-81.
- (69) Boemers TM, Beek F J A, Bax N M A. Guidelines for the urological screening and initial management of lower urinary tract dysfunction in children with anorectal malformations - the ARGUS protocol. *BJU International* 1999;83:662-71.
- (70) Patwardhan N, Kiely EM, Drake DP, Spitz L, Pierro A. Colostomy for anorectal anomalies: high incidence of complications. *J Pediatr Surg* 2001 May;36(5):795-8.
- (71) Cigdem MK, Onen A, Duran H, Ozturk H, Otcu S. The mechanical complications of colostomy in infants and children: analysis of 473 cases of a single center. *Pediatr Surg Int* 2006 Aug;22(8):671-6.

- (72) deVries PA, Pena A. Posterior sagittal anorectoplasty. *J Pediatr Surg* 1982 Oct;17(5):638-43.
- (73) Pena A. Male Defects. Atlas of surgical management of anorectal malformations. New York: Springer-Verlag; 1989. p. 26-44.
- (74) Moore TC. Advantages of performing the sagittal anoplasty operation for imperforate anus at birth. *J Pediatr Surg* 1990 Feb;25(2):276-7.
- (75) Adeniran JO. One-stage correction of imperforate anus and rectovestibular fistula in girls: Preliminary results. *J Pediatr Surg* 2002 Jun;37(6):E16.
- (76) Liu G, Yuan J, Geng J, Wang C, Li T. The treatment of high and intermediate anorectal malformations: one stage or three procedures? *J Pediatr Surg* 2004 Oct;39(10):1466-71.
- (77) Georgeson KE, Inge TH, Albanese CT. Laparoscopically assisted anorectal pull-through for high imperforate anus--a new technique. *J Pediatr Surg* 2000 Jun;35(6):927-30.
- (78) Tei E, Yamataka A, Segawa O, Kobayashi H, Lane GJ, Tobayama S, et al. Laparoscopically assisted anorectovaginoplasty for selected types of female anorectal malformations. *J Pediatr Surg* 2003 Dec;38(12):1770-4.
- (79) Pena A, Guardino K, Tovilla JM, Levitt MA, Rodriguez G, Torres R. Bowel management for fecal incontinence in patients with anorectal malformations. *J Pediatr Surg* 1998 Jan;33(1):133-7.
- (80) Pena A. Continence after posterior sagittal anorectoplasty. *J Pediatr Surg* 1992 Mar;27(3):415-7.
- (81) Pena A, el Behery M. Megacigmoid: a source of pseudoincontinence in children with repaired anorectal malformations. *J Pediatr Surg* 1993 Feb;28(2):199-203.
- (82) Malone PS, Ransley PG, Kiely EM. Preliminary report: the antegrade continence enema. *Lancet* 1990 Nov 17;336(8725):1217-8.
- (83) Shandling B, Chait PG, Richards HF. Percutaneous cecostomy: a new technique in the management of fecal incontinence. *J Pediatr Surg* 1996 Apr;31(4):534-7.
- (84) Monti PR, Lara RC, Dutra MA, de C, Jr. New techniques for construction of efferent conduits based on the Mitrofanoff principle. *Urology* 1997 Jan;49(1):112-5.
- (85) Liloku RB, Mure PY, Braga L, Basset T, Mouriquand PD. The left Monti-Malone procedure: Preliminary results in seven cases. *J Pediatr Surg* 2002 Feb;37(2):228-31.

- (86) Kim J, Beasley SW, Maoate K. Appendicostomy stomas and antegrade colonic irrigation after laparoscopic antegrade continence enema. *J Laparoendosc Adv Surg Tech A* 2006 Aug;16(4):400-3.
- (87) Ameda K, Kakizaki H, Machino R, Tanaka H, Shibata T, Koyanagi T. Laparoscopic antegrade continence enema procedure for fecal incontinence in a patient with spina bifida. *Int J Urol* 2003 Jul;10(7):401-3.
- (88) Levitt MA, Soffer SZ, Pena A. Continent appendicostomy in the bowel management of fecally incontinent children. *J Pediatr Surg* 1997 Nov;32(11):1630-3.
- (89) Ellsworth PI, Webb HW, Crump JM, Barraza MA, Stevens PS, Mesrobian HGJ. The Malone Antegrade Colonic Enema Enhances the Quality of Life in Children Undergoing Urological Incontinence Procedures. *J Urol* 1996;155(4):1416-8.
- (90) Yerkes EB, Cain MP, King S, Brei T, Kaefer M, Casale AJ, et al. The Malone antegrade continence enema procedure: quality of life and family perspective. *J Urol* 2003 Jan;169(1):320-3.
- (91) Dey R, Ferguson C, Kenny SE, Shankar KR, Coldicutt P, Baillie CT, et al. After the honeymoon--medium-term outcome of antegrade continence enema procedure. *J Pediatr Surg* 2003 Jan;38(1):65-8.
- (92) Curry JJ, Osborne A, Malone PS. How to achieve a successful Malone antegrade continence enema. *J Pediatr Surg* 1998 Jan;33(1):138-41.
- (93) PICKRELL KL, BROADBENT TR, MASTERS FW, METZGER JT. Construction of a rectal sphincter and restoration of anal continence by transplanting the gracilis muscle; a report of four cases in children. *Ann Surg* 1952 Jun;135(6):853-62.
- (94) Kotobi H, Forin V, Larroquet M, Khairouni A, Loc'h P, Grapin C, et al. [Pickrell intervention in children for anal incontinence secondary to anorectal malformation]. *Ann Chir* 2000 Dec;125(10):954-60.
- (95) PICKRELL KL, BROADBENT TR, MASTERS FW, METZGER JT. Construction of a rectal sphincter and restoration of anal continence by transplanting the gracilis muscle; a report of four cases in children. *Ann Surg* 1952 Jun;135(6):853-62.
- (96) Baeten CG, Uludag OO, Rongen MJ. Dynamic graciloplasty for fecal incontinence. *Microsurgery* 2001;21(6):230-4.
- (97) Koch SM, Uludag O, Rongen MJ, Baeten CG, van GW. Dynamic graciloplasty in patients born with an anorectal malformation. *Dis Colon Rectum* 2004 Oct;47(10):1711-9.
- (98) Christiansen J, Lorentzen M. Implantation of artificial sphincter for anal incontinence. *Lancet* 1987 Aug 1;2(8553):244-5.

- (99) Altomare DF, Dodi G, La TF, Romano G, Melega E, Rinaldi M. Multicentre retrospective analysis of the outcome of artificial anal sphincter implantation for severe faecal incontinence. *Br J Surg* 2001 Nov;88(11):1481-6.
- (100) Vaizey CJ, Kamm MA, Gold DM, Bartram CI, Halligan S, Nicholls RJ. Clinical, physiological, and radiological study of a new purpose-designed artificial bowel sphincter. *Lancet* 1998 Jul 11;352(9122):105-9.
- (101) da Silva GM, Jorge JM, Belin B, Noguerras JJ, Weiss EG, Vernava AM, et al. New Surgical Options for Fecal Incontinence in Patients With Imperforate Anus. *Diseases of the Colon & Rectum* 2004 Feb;47(2):204-9.
- (102) Baird AD, Mathews RI, Gearhart JP. The use of combined bladder and epispadias repair in boys with classic bladder exstrophy: outcomes, complications and consequences. *J Urol* 2005 Oct;174(4 Pt 1):1421-4.
- (103) Sheldon CA, Gilbert A, Lewis AG, Aiken J, Ziegler MM. Surgical Implications of Genitourinary Tract Anomalies in Patients with Imperforate Anus. *J Urol* 1993;152:196-9.
- (104) Gilbert SM, Hensle TW. Metabolic Consequences and Long-term Complications of Enterocystoplasty in Children: A Review. *J Urol* 2005;173:1080-6.
- (105) Gunaydin M, Ariturk E, Rizalar R, Gork S, Bernay F, Gurses N. Unilateral inguinal ectopic scrotum and imperforate anus: a case report. *J Pediatr Surg* 1997 Sep;32(9):1360-1.
- (106) Farnham SB, Cookson MS. Surgical Complications of Urinary Diversion. *World J Urol* 2004;X:X.
- (107) Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW, III. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. *J Urol* 1997 Sep;158(3 Pt 2):1298-300.
- (108) Woodhouse CR. The sexual and reproductive consequences of congenital genitourinary anomalies. *J Urol* 1994 Aug;152(2 Pt 2):645-51.
- (109) Gasquet I, Falissard B, Ravaud P. Impact of reminders and method of questionnaire distribution on patient response to mail-back satisfaction survey. *J Clin Epidemiol* 2001 Nov;54(11):1174-80.
- (110) Perneger TV, Chamot E, Bovier PA. Nonresponse bias in a survey of patient perceptions of hospital care. *Med Care* 2005 Apr;43(4):374-80.
- (111) Hassink EA, Rieu PN, Brugman AT, Festen C. Quality of life after operatively corrected high anorectal malformation: a long-term follow-up study of patients aged 18 years and older. *J Pediatr Surg* 1994 Jun;29(6):773-6.
- (112) Kelly JH. Cine radiography in anorectal malformations. *Journal of Pediatric Surgery* 1969 Oct;4(5):538-46.

- (113) Rintala RJ, Lindahl H. Is normal bowel function possible after repair of intermediate and high anorectal malformations? *J Pediatr Surg* 1995 Mar;30(3):491-4.
- (114) Doolin EJ, Black CT, Donaldson JS, Schwartz D, Raffensperger JG. Rectal manometry, computed tomography, and functional results of anal atresia surgery. *J Pediatr Surg* 1993 Feb;28(2):195-8.
- (115) Taylor I, Duthie HL, Zachary RB. Anal continence following surgery for imperforate anus. *J Pediatr Surg* 1973 Aug;8(4):497-503.
- (116) Pescatori M, Anastasio G, Bottini C, Mentasti A. New grading and scoring for anal incontinence. Evaluation of 335 patients. *Dis Colon Rectum* 1992 May;35(5):482-7.
- (117) American Medical Systems. Fecal incontinence scoring system. Minnetonka, American Medical Systems.
Ref Type: Generic
- (118) Jorge JM, Wexner SD. Etiology and management of fecal incontinence. *Dis Colon Rectum* 1993 Jan;36(1):77-97.
- (119) Vaizey CJ, Carapeti E, Cahill JA, Kamm MA. Prospective comparison of faecal incontinence grading systems. *Gut* 1999 Jan;44(1):77-80.
- (120) Bliss DP, Jr., Tapper D, Anderson JM, Schaller RT, Jr., Hatch EI, Morgan A, et al. Does posterior sagittal anorectoplasty in patients with high imperforate anus provide superior fecal continence? *J Pediatr Surg* 1996 Jan;31(1):26-30.
- (121) Holschneider AM, Pfrommer W, Gerresheim B. Results in the treatment of anorectal malformations with special regard to the histology of the rectal pouch. *Eur J Pediatr Surg* 1994 Oct;4(5):303-9.
- (122) Agachan F, Chen T, Pfeifer J, Reissman P, Wexner SD. A constipation scoring system to simplify evaluation and management of constipated patients. *Dis Colon Rectum* 1996 Jun;39(6):681-5.
- (123) McLorie GA, Sheldon CA, Fleisher M, Churchill BM. The genitourinary system in patients with imperforate anus. *J Pediatr Surg* 1987 Dec;22(12):1100-4.
- (124) Munn R, Schillinger JF. Urologic abnormalities found with imperforate anus. *Urology* 1983 Mar;21(3):260-4.
- (125) Hoekstra WJ, Scholtmeijer RJ, Molenaar JC, Schreeve RH, Schroeder FH. Urogenital tract abnormalities associated with congenital anorectal anomalies. *J Urol* 1983 Nov;130(5):962-3.
- (126) Misra D, Mushtaq I, Drake DP, Kiely EM, Spitz L. Associated urologic anomalies in low imperforate anus are capable of causing significant morbidity: a 15-year experience. *Urology* 1996 Aug;48(2):281-3.

- (127) Jackson S, Donovan J, Brookes S, Eckford S, Swithinbank L, Abrams P. The Bristol Female Lower Urinary Tract Symptoms questionnaire: development and psychometric testing. *BJU Int* 1996;77:805-12.
- (128) Edwards P, Roberts I, Clarke M, DiGuseppi C, Prata S, Wentz R, et al. Increasing response rates to postal questionnaires: systematic review. *BMJ* 2002 May 18;324:1183-6.
- (129) Donovan, Brookes, De La R, Peters, Porru, Kondo, et al. The responsiveness of the ICSmale questionnaire to outcome: evidence from the ICS-'BPH' study. *BJU International* 1999 Feb 1;83(3):243-8.
- (130) World Health Organisation. Constitution. Chronical 1. 1947. Geneva, World Health Organisation.
Ref Type: Generic
- (131) Browne JP. Health-related quality-of-life studies in urology: conceptual and methodological considerations. *World J Urol* 1999 Aug;17(4):193-8.
- (132) Ware JE, Jr., Sherbourne CD. The MOS 36-item short-form health survey (SF-36). I. Conceptual framework and item selection. *Med Care* 1992 Jun;30(6):473-83.
- (133) Brazier JE, Harper R, Jones NM, O'Cathain A, Thomas KJ, Usherwood T, et al. Validating the SF-36 health survey questionnaire: new outcome measure for primary care. *BMJ* 1992 Jul 18;305(6846):160-4.
- (134) World Health Organisation. Education and treatment in human sexuality: The training of health professionals. Geneva: WHO; 1975.
- (135) Snell WE, Fisher TD, Walters AS. The Multidimensional Sexuality Questionnaire: An objective self-report measure of psychological tendencies associated with human sexuality. *Annals of Sex Research* 1993;6:27-55.
- (136) O'Leary MP, Fowler FJ, Lenderking WR, Barber B, Sagnier PP, Guess HA, et al. A brief male sexual function inventory for urology. *Urology* 1995 Nov;46(5):697-706.
- (137) Mayer JD, Eisenberg MG. Mental representations of the body: Stability and change in response to illness and disability. *Rehabil Psychol* 1988;33:155-71.
- (138) Lawrence B. Self-concept formation and physical handicap: some educational implication for intergration. *Disabil Handicap Soc* 1991;6:139-46.
- (139) Franzoi SL, Shields SA. The Body Esteem Scale: multidimensional structure and sex differences in a college population. *J Pers Assess* 1984 Apr;48(2):173-8.
- (140) Franzoi SL. Further evidence of the reliability and validity of the Body Esteem Scale. *J Clin Psychol* 1994 Mar;50(2):237-9.

- (141) Rhodes T, Girman CJ, Jacobsen SJ, Guess HA, Hanson KA, Oesterling JE, et al. Does the mode of questionnaire administration affect the reporting of urinary symptoms? *Urology* 1995 Sep;46(3):341-5.
- (142) Rodriguez LV, Blander DS, Dorey F, Raz S, Zimmern P. Discrepancy in patient and physician perception of patient's quality of life related to urinary symptoms. *Urology* 2003;62(1):49-53.
- (143) Edwards P, Roberts I, Clarke M, DiGuseppi C, Pratap S, Wentz R, et al. Increasing response rates to postal questionnaires: systematic review. *BMJ* 2002 May 18;324(7347):1183.
- (144) Kelly JH. Cine radiography in anorectal malformations. *Journal of Pediatric Surgery* 1969 Oct;4(5):538-46.
- (145) Lyons RA, Wareham K, Lucas M, Price D, Williams J, Hutchings HA. SF-36 scores vary by method of administration: implications for study design. *J Public Health Med* 1999 Mar;21(1):41-5.
- (146) Meyer KF, Freitas Filho LG, Martins DM, Vaccari M, Carnevale J. The exstrophy-epispadias complex: is aesthetic appearance important? *BJU Int* 2004 May;93(7):1062-8.
- (147) Woodhouse CR. The sexual and reproductive consequences of congenital genitourinary anomalies. *J Urol* 1994 Aug;152(2 Pt 2):645-51.
- (148) Cervino AC, Hill AV. Comparison of tests for association and linkage in incomplete families. *Am J Hum Genet* 2000 Jul;67(1):120-32.
- (149) Census. Office for National Statistics; 2001.
- (150) NRS Ltd. National Readership Survey: Jan-Dec. 2005.
- (151) Temtamy SA, Miller JD. Extending the scope of the VATER association: one end of a spectrum of anomalies. *J Pediatr* 1974;85:345.
- (152) Cuschieri A, EUROCAT Working Group. Anorectal Anomalies Associated With or as Part of Other Anomalies. *American Journal of Medical Genetics* 2002 Feb;110:122-30.
- (153) Ratan SK, Rattan KN, Pandey RM, Mittal A, Magu S, Sodhi PK. Associated congenital anomalies in patients with anorectal malformations--a need for developing a uniform practical approach. *J Pediatr Surg* 2004 Nov;39(11):1706-11.
- (154) Malone PS. The management of bowel problems in children with urological disease. *Br J Urol* 1995 Aug;76(2):220-5.
- (155) Jorge JM, Wexner SD. Etiology and management of fecal incontinence. *Dis Colon Rectum* 1993 Jan;36(1):77-97.
- (156) Pena A. Anorectal malformations. *Semin Pediatr Surg* 1995 Feb;4(1):35-47.

- (157) Diseth TH, Emblem R. Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. *J Pediatr Surg* 1996 May;31(5):638-43.
- (158) Karkowski JK, Pollock WF, Landon CW. Imperforate anus. Eighteen to thirty year follow-up study. *Am J Surg* 1973 Aug;126(2):141-7.
- (159) Nixon HH, Puri P. The results of treatment of anorectal anomalies: a thirteen to twenty year follow-up. *J Pediatr Surg* 1977 Feb;12(1):27-37.
- (160) Rintala R, Mildh L, Lindahl H. Fecal continence and quality of life in adult patients with an operated low anorectal malformation. *J Pediatr Surg* 1992 Jul;27(7):902-5.
- (161) Hassink EA, Rieu PN, Severijnen RS, vd Staak FH, Festen C. Are adults content or continent after repair for high anal atresia? A long-term follow-up study in patients 18 years of age and older. *Ann Surg* 1993 Aug;218(2):196-200.
- (162) Rintala R, Mildh L, Lindahl H. Fecal continence and quality of life for adult patients with an operated high or intermediate anorectal malformation. *J Pediatr Surg* 1994 Jun;29(6):777-80.
- (163) Nixon HH, Puri P. The results of treatment of anorectal anomalies: a thirteen to twenty year follow-up. *J Pediatr Surg* 1977 Feb;12(1):27-37.
- (164) Rintala R, Mildh L, Lindahl H. Fecal continence and quality of life for adult patients with an operated high or intermediate anorectal malformation. *J Pediatr Surg* 1994 Jun;29(6):777-80.
- (165) Hamid CH, Holland AJ, Martin HC. Long-term outcome of anorectal malformations: the patient perspective. *Pediatr Surg Int* 2007 Feb;23(2):97-102.
- (166) Pena A, Hong A. Advances in the management of anorectal malformations. *Am J Surg* 2000 Nov;180(5):370-6.
- (167) Rintala RJ, Lindahl HG. Fecal continence in patients having undergone posterior sagittal anorectoplasty procedure for a high anorectal malformation improves at adolescence, as constipation disappears. *J Pediatr Surg* 2001 Aug;36(8):1218-21.
- (168) Agachan F, Chen T, Pfeifer J, Reissman P, Wexner SD. A constipation scoring system to simplify evaluation and management of constipated patients. *Dis Colon Rectum* 1996 Jun;39(6):681-5.
- (169) Saunders JR, Williams NS, Eccersley AJ. The combination of electrically stimulated gracilis neoanal sphincter and continent colonic conduit: a step forward for total anorectal reconstruction? *Dis Colon Rectum* 2004 Mar;47(3):354-63.

- (170) da Silva GM, Jorge JM, Belin B, Nogueras JJ, Weiss EG, Vernava AM, et al. New Surgical Options for Fecal Incontinence in Patients With Imperforate Anus. *Diseases of the Colon & Rectum* 2004 Feb;47(2):204-9.
- (171) Donovan, Brookes, De La R, Peters, Porru, Kondo, et al. The responsiveness of the ICSmale questionnaire to outcome: evidence from the ICS-'BPH' study. *BJU International* 1999 Feb 1;83(3):243-8.
- (172) Misra D, Mushtaq I, Drake DP, Kiely EM, Spitz L. Associated urologic anomalies in low imperforate anus are capable of causing significant morbidity: a 15-year experience. *Urology* 1996 Aug;48(2):281-3.
- (173) van DM, Skrekas T, de la Rosette JJ. The association between lower urinary tract symptoms and sexual dysfunction: fact or fiction? *Curr Opin Urol* 2005 Jan;15(1):39-44.
- (174) Coyne KS, Zhou Z, Thompson C, Versi E. The impact on health-related quality of life of stress, urge and mixed urinary incontinence. *BJU International* 2003;92:731-5.
- (175) Terai A, Matsui Y, Ichioka K, Ohara H, Terada N, Yoshimura K. Comparative Analysis of Lower Urinary Tract Symptoms and Bother in Both Sexes. *Urology* 2004;63(3):487-91.
- (176) Davies MC, Crouch NS, Woodhouse CR, Creighton SM. Congenital adrenal hyperplasia and lower urinary tract symptoms. *BJU Int* 2005 Jun;95(9):1263-6.
- (177) Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW, III. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. *J Urol* 1997 Sep;158(3 Pt 2):1298-300.
- (178) McLorie GA, Sheldon CA, Fleisher M, Churchill BM. The genitourinary system in patients with imperforate anus. *J Pediatr Surg* 1987 Dec;22(12):1100-4.
- (179) Boemers TM, de Jong TP, van Gool JD, Bax KM. Urologic problems in anorectal malformations. Part 2: functional urologic sequelae. *J Pediatr Surg* 1996 May;31(5):634-7.
- (180) Pena A. Anorectal Malformations: Experience with the Posterior Sagittal Approach. In: Mark D, Oldham K T, Mouriquand P, Edward R, editors. *Pediatric Surgery and Urology: Long Term Outcomes*. 1 ed. Saunders; 1998. p. 376-86.
- (181) Rhodes T, Girman CJ, Jacobsen SJ, Guess HA, Hanson KA, Oesterling JE, et al. Does the mode of questionnaire administration affect the reporting of urinary symptoms? *Urology* 1995 Sep;46(3):341-5.
- (182) Ralph DJ, Woodhouse CR, Ransley PG. The management of the neuropathic bladder in adolescents with imperforate anus. *J Urol* 1992 Aug;148(2 Pt 1):366-8.

- (183) Tank ES, Erns CB, Woolson ST. Urinary tract complications of anorectal surgery. *Am J Surg* 1972;123:118-22.
- (184) Wiener ES, Kieseewetter WB. Urologic abnormalities associated with imperforate anus. *J Pediatr Surg* 1973;8:151-7.
- (185) Boemers TM, Bax KM, Rovekamp MH, van Gool JD. The effect of posterior sagittal anorectoplasty and its variants on lower urinary tract function in children with anorectal malformations. *J Urol* 1995 Jan;153(1):191-3.
- (186) Woodhouse CR. The fate of the abnormal bladder in adolescence. *J Urol* 2001 Dec;166(6):2396-400.
- (187) O'Leary MP, Fowler FJ, Lenderking WR, Barber B, Sagnier PP, Guess HA, et al. A brief male sexual function inventory for urology. *Urology* 1995 Nov;46(5):697-706.
- (188) O'Leary MP, Rhodes T, Girman CJ, Jacobson DJ, Roberts RO, Lieber MM, et al. Distribution of the Brief Male Sexual Inventory in community men. *Int J Impot Res* 2003 Jun;15(3):185-91.
- (189) Patankar JZ, Mali VP, Yashpal R, Neo GT, Prabhakaran K. Anorectal malformation with congenital absence of vagina: a case report and review of the literature. *Pediatr Surg Int* 2004 Apr;20(4):295-7.
- (190) Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW, III. Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. *J Urol* 1997 Sep;158(3 Pt 2):1298-300.
- (191) Warne SA, Wilcox DT, Creighton S, Ransley PG. Long-term gynecological outcome of patients with persistent cloaca. *J Urol* 2003 Oct;170(4 Pt 2):1493-6.
- (192) Levitt MA, Stein DM, Pena A. Gynecologic concerns in the treatment of teenagers with cloaca. *J Pediatr Surg* 1998 Feb;33(2):188-93.
- (193) Hoekstra WJ, Scholtmeijer RJ, Molenaar JC, Schreeve RH, Schroeder FH. Urogenital tract abnormalities associated with congenital anorectal anomalies. *Journal of Urology* 1983;130:962-3.
- (194) Cho S, Moore SP, Fangman T. One hundred three consecutive patients with anorectal malformations and their associated anomalies. *Arch Pediatr Adolesc Med* 2001 May;155(5):587-91.
- (195) Spouge D, Baird PA. Imperforate anus in 700,000 consecutive liveborn infants. *Am J Med Genet Suppl* 1986;2:151-61.
- (196) Cortes D, Thorup JM, Nielsen OH, Beck BL. Cryptorchidism in boys with imperforate anus. *J Pediatr Surg* 1995 Apr;30(4):631-5.

- (197) Bianchini MA, Fava G, Cortese MG, Vinardi S, Costantino S, Canavese F. A rare anorectal malformation: a very large H-type fistula. *Pediatr Surg Int* 2001 Nov;17(8):649-51.
- (198) Woodhouse CR. Prospects for fertility in patients born with genitourinary anomalies. *J Urol* 2001 Jun;165(6 Pt 2):2354-60.
- (199) Holt B, Pryor JP, Hendry WF. Male infertility after surgery for imperforate anus. *J Pediatr Surg* 1995 Dec;30(12):1677-9.
- (200) Salonia A, Zanni G, Nappi R, Briganti A, Deho F, Fabbri F, et al. Sexual dysfunction is common in women with lower urinary tract symptoms and urinary incontinence: Results of a cross-sectional study. *Eur Urol* 2004;45:642-8.
- (201) Rintala R. Anorectal malformations: an overview. In: Stringer MD, Mouriquand PDE, Oldham KT, Howard ER, editors. *Pediatric Surgery and Urology: Longterm Outcomes*. 1 ed. London: WBSaunders; 1998. p. 357-75.
- (202) Woodhouse CRJ. Myelomeningocele in young adults. *British Journal of Urology International* 2005;95:223-30.
- (203) Holt B, Pryor JP, Hendry WF. Male infertility after surgery for imperforate anus. *J Pediatr Surg* 1995 Dec;30(12):1677-9.
- (204) Shiraishi K, Takihara H. Recurrent epididymo-orchitis caused by posterior urethral valve associated with imperforate anus. *Int J Urol* 2004 Jan;11(1):58-60.
- (205) Megalli M, Gursel E, Lattimer JK. Reflux of urine into ejaculatory ducts as a cause of recurring epididymitis in children. *J Urol* 1972 Dec;108(6):978-9.
- (206) Oguzkurt P, Tanyel FC, Buyukpamukcu N. Acute scrotum due to edidymo-orchitis associated with vasal anomalies in children with anorectal malformations. *J Pediatr Surg* 1998 Dec;33(12):1834-6.
- (207) Schober JM. Feminising genitoplasty for intersex. In: Stringer MD, Oldham KT, Mouriquand PDE, Howard ER, editors. *Paediatric Surgery and Urology: Long Term Outcomes*. 1 ed. London: W.B.Saunders; 1999. p. 549-58.
- (208) Steiner E, Woernle F, Kuhn W, Beckmann K, Schmidt M, Pilch H, et al. Carcinoma of the neovagina: case report and review of the literature. *Gynecol Oncol* 2002 Jan;84(1):171-5.
- (209) Wellings K, Nanchahal K, Macdowall W, McManus S, Erens B, Mercer CH, et al. Sexual behaviour in Britain: early heterosexual experience. *Lancet* 2001 Dec 1;358(9296):1843-50.
- (210) Keles H, Ekici A, Ekici M, Bulcun E, Altinkaya V. Effect of chronic diseases and associated psychological distress on health-related quality of life. *Intern Med J* 2007 Jan;37(1):6-11.

- (211) Ludman L. Anorectal Malformations - Psychological aspects. In: Stringer M, Oldham K T, Mouriquand P, Howard E, editors. *Pediatric Surgery & Urology: Long-term Outcomes*. 1st ed. London: WB Saunders; 1998. p. 387-93.
- (212) Hassink EA, Brugman-Boezeman AT, Robbroeckx LM, Rieu PN, van Kuyk EM, Wels PM, et al. Parenting children with anorectal malformations: implications and experiences. *Pediatr Surg Int* 1998 Jul;13(5-6):377-83.
- (213) Diseth TH, Emblem R. Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. *J Pediatr Surg* 1996 May;31(5):638-43.
- (214) Ludman L, Spitz L, Tsuji H, Pierro A. Hirschsprung's disease: functional and psychological follow up comparing total colonic and rectosigmoid aganglionosis. *Arch Dis Child* 2002 May;86(5):348-51.
- (215) Bai Y, Yuan Z, Wang W, Zhao Y, Wang H, Wang W. Quality of life for children with fecal incontinence after surgically corrected anorectal malformation. *J Pediatr Surg* 2000 Mar;35(3):462-4.
- (216) Iwai N, Deguchi E, Kimura O, Kubota Y, Ono S, Shimadera S. Social quality of life for adult patients with anorectal malformations. *J Pediatr Surg* 2007 Feb;42(2):313-7.
- (217) Franzoi SL, Shields SA. The Body Esteem Scale: multidimensional structure and sex differences in a college population. *J Pers Assess* 1984 Apr;48(2):173-8.
- (218) Franzoi SL. Further evidence of the reliability and validity of the Body Esteem Scale. *J Clin Psychol* 1994 Mar;50(2):237-9.
- (219) Franzoi SL, Shields SA. The Body Esteem Scale: multidimensional structure and sex differences in a college population. *J Pers Assess* 1984 Apr;48(2):173-8.
- (220) Diseth TH, Emblem R. Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. *J Pediatr Surg* 1996 May;31(5):638-43.
- (221) Aminoff D, La Sala E, Zaccara A. Follow-up of anorectal anomalies: the Italian parents' and patients' perspective. *J Pediatr Surg* 2006;41:837-41.
- (222) von RU, Gosch A, Rajmil L, Bisegger C, Ravens-Sieberer U. Socioeconomic determinants of health related quality of life in childhood and adolescence: results from a European study. *J Epidemiol Community Health* 2006 Feb;60(2):130-5.
- (223) McLaren L, Kuh D. Women's body dissatisfaction, social class, and social mobility. *Soc Sci Med* 2004;58(9):1575-84.

- (224) Paeratakul S, White MA, Williamson DA, Ryan DH, Bray GA. Sex, race/ethnicity, socioeconomic status, and BMI in relation to self-perception of overweight. *Obes Res* 2002 May;10(5):345-50.
- (225) Hassink EA, Rieu PN, Brugman AT, Festen C. Quality of life after operatively corrected high anorectal malformation: a long-term follow-up study of patients aged 18 years and older. *J Pediatr Surg* 1994 Jun;29(6):773-6.
- (226) Hamid CH, Holland AJ, Martin HC. Long-term outcome of anorectal malformations: the patient perspective. *Pediatr Surg Int* 2007 Feb;23(2):97-102.
- (227) Pena A, Hong A. Advances in the management of anorectal malformations. *Am J Surg* 2000 Nov;180(5):370-6.
- (228) Woodhouse CR. The fate of the abnormal bladder in adolescence. *J Urol* 2001 Dec;166(6):2396-400.
- (229) Office for National Statistics. Birth Statistics. Office for National Statistics 2006 Available from: URL: <http://www.statistics.gov.uk/>
- (230) Office for National Statistics. Psychiatric Morbidity Report. 2001.
- (231) Jenetzky EWD, Schwazer N. ARM: Aftercare and Impact from the Perspective of the Family. In: Holschneider AM, Hutson JM, editors. *Anorectal Malformations in Children: Embryology, Diagnosis, Surgical Treatment, Follow-up*. Berlin Heidelberg New York: Springer; 2006. p. 459-69.

Appendix 1

ACADEMIC DEPARTMENT OF OBSTETRICS & GYNAECOLOGY
University College London
86-96 Chenies Mews
London WC1E 6HX

Tel: 020 7679 6654
Fax: 020 7383 7429

Head of Department Professor CH Rodeck DSc, FRCOG, FRCPath



Dear

We are currently performing a study looking at the outcome in adults born with certain conditions, and we would be grateful if you would be prepared to help with our study.

Participation in the study would involve filling in questionnaires about your general health, the surgery you have had, questions about your bowel and urinary function, and some more personal questions about your personal and sex life. These questionnaires are strictly confidential and the information is treated in an anonymous way, so that you are not personally identifiable.

The second part of the study is an invitation to visit us in a clinic in London for an examination. This is not compulsory and you can participate with as much as you feel comfortable with. The travel costs you incur will be reimbursed.

All data collected will be stored in a secure manner according to the Data Protection Act 1998, and UCL Hospital guidelines. We aim to publish our results in the medical literature to improve doctors and other health professionals understanding of a complicated condition.

We would be grateful if you could fill in the form enclosed to let us know if you would like to participate or not and return it in the addressed envelope.

Many thanks.

Yours sincerely,

Dr Melissa Davies
Research Fellow
University College London

Mr Duncan Wilcox
Consultant Paediatric Urologist
Great Ormond Street Hospital

Miss Sarah Creighton
Consultant Gynaecologist
University College Hospital

Appendix 2

ACADEMIC DEPARTMENT OF OBSTETRICS & GYNAECOLOGY
University College London
86-96 Chenies Mews
London WC1E 6HX

Tel: 020 7679 6654
Fax: 020 7383 7429

Head of Department Professor CH Rodeck DSc, FRCOG, FRCPath



Patient Information Sheet – Jan 2004

*** CONFIDENTIAL ***

Long Term Outcomes of Cloacal and Anorectal Malformations

You are being invited to take part in a research study. Before you decide whether to take part it is important for you to understand what it will involve. Ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

Anorectal and cloacal anomalies are rare conditions which involve abnormal development of the genital area. Patients with these conditions usually need to have complex operations as babies and small children in order for the bowel and bladder to work as well as possible.

It is known that the genital anatomy is closely linked with the development of the bladder and the bowel. This may mean that sometimes there is an unusual development of the reproductive or sexual apparatus.

In the past children with these conditions have been unwell and were not expected to survive into adulthood. In the last 30 years there have been major improvements in surgical and intensive care techniques. This means that people born with these conditions can expect to lead relatively normal lives, which includes having a sex life, and having children.

However, it is not known if there are any problems with this. There have been isolated reports of females who have undergone corrective treatment getting pregnant and having children, but there are no large studies looking at this. There are also no studies looking at whether patients with this condition have any problems with sexual intercourse, or relationships.

We would like to find out how patients with these conditions are in terms of their health and quality of life, once they have become adults. This information would be

useful to give parents of babies born with this condition today, and for the paediatric surgeons operating on these patients.

Our study has two parts, the first part involves a collection of questionnaires looking at general health, past medical and surgical details, how you feel about yourself, your personal life and any difficulties you may have experienced in these areas. The second part is to come to clinic to meet me for a discussion and an examination. The general examination involves looking at your abdomen (tummy), and looking at your genital area and performing some gentle tests. You may choose to participate in as much or as little as you feel comfortable with.

All of the information that we collect is strictly confidential. It is stored in a locked cabinet, with only the study investigator having access to it. Any data stored on a computer is anonymous, and is password protected, in accordance with the Data Protection Act 1998, and with UCL hospital guidelines. None of this information is passed onto other hospitals, doctors, or organisations. We aim to publish our findings in the medical literature to inform doctors, other health care professionals and patients/parents. **There will be nothing which could lead to your identification.**

If you decide to take part you are free to withdraw at any time and without giving a reason. A decision to withdraw at any time or a decision not to take part will not affect the standard of future care you may receive.

Thank you for taking the time to read this information sheet.

Dr Melissa Davies
Clinical Research Fellow
University College London

Mr Duncan Wilcox
Consultant Paediatric Urologist
Great Ormond Street Hospital

Miss Sarah Creighton
Consultant Gynaecologist
University College Hospital

Appendix 3

Survey into Adult Outcomes after
Anorectal Malformation and Cloaca
Repair.



CONFIDENTIAL

ACADEMIC DEPARTMENT OF OBSTETRICS & GYNAECOLOGY
University College London
86-96 Chenies Mews
London WC1E 6HX

Tel: 020 7679 6654
Fax: 020 7383 7429

**Head of Department Professor CH Rodeck DSc, FRCOG,
FRCPath**



UCLH Project ID - 02/0287
Patient Identification Number:

CONSENT FORM

Survey into Adult Outcomes after Anorectal Malformation and Cloaca Repair.

Please read the information sheet.

Thank you for agreeing to take part in our study, please could you complete the questionnaires and return them in the stamped addressed envelope enclosed. If you have any queries relating to the study or any difficulties with the questionnaires please contact Dr Melissa Davies on the details above.

Please read and delete as appropriate:

Have you read the information about this study ?	YES/NO
Have you received enough information about this study?	YES/NO

Do you understand that you are free to withdraw from this study

- | | |
|---|--------|
| <ul style="list-style-type: none">▪ at any time▪ without having to give a reason▪ without affecting your future medical care? | YES/NO |
|---|--------|

Do you agree to take part in this study?	YES/NO
--	--------

Name of patient

Date

Signature

If you have any comments or concerns you may discuss these with the investigator. If you wish to go further and complain about any aspect of the way you have been approached or treated during the course of the study, you should write or get in touch with the Complaints Manager, UCL hospitals. Please quote the UCLH project number at the top of this consent form.

ACADEMIC DEPARTMENT OF OBSTETRICS & GYNAECOLOGY
University College London
86-96 Chenies Mews
London WC1E 6HX

Tel: 020 7679 6654
Fax: 020 7383 7429

Head of Department Professor CH Rodeck DSc, FRCOG, FRCPath



Access to Medical Notes

We would like to be able to view your medical records from other hospitals where you have received treatment. This is gain a complete record of all operations, investigations and treatment you have had.

To do this we would like your permission to be able to look at your notes from other hospitals where you have been a patient.

If you agree to this please complete the form below.

Full Name (current)
Previous names used
Date of Birth
Current Address

.....

Hospitals where you have been treated

.....
.....
.....
.....
.....

I give permission for Dr Melissa Davies of University College London to view my hospital case notes

Signature.....

Date.....

Social and Demographic Information

This section tells us whether our survey sample is similar to the UK population in social and economic terms. The information is not for identifying you.

Your age:

Your gender: Male / Female

Which best describes your ethnic background?

- | | | | |
|--------------------------------------|--|--|--|
| <input type="checkbox"/> Bangladeshi | <input type="checkbox"/> Black African | <input type="checkbox"/> Black British | <input type="checkbox"/> Black Caribbean |
| <input type="checkbox"/> Black other | <input type="checkbox"/> British Asian | <input type="checkbox"/> Chinese | <input type="checkbox"/> Indian |
| <input type="checkbox"/> Pakistani | <input type="checkbox"/> White | <input type="checkbox"/> | |
- Other: _____

What is the highest qualification that you have passed?

- | | |
|--|---|
| <input type="checkbox"/> Degree or equivalent | <input type="checkbox"/> Non-degree teaching or other qualification |
| <input type="checkbox"/> 'A' level or equivalent | <input type="checkbox"/> GCSE, 'O' level or equivalent |
| <input type="checkbox"/> GCSE or equivalent | <input type="checkbox"/> Other qualifications <input type="checkbox"/> No qualification |

Are you currently in a relationship? (Tick one box only of six)

- YES: ☐ Married, living together ☐ Not married, living together ☐ Not living together
- NO: ☐ Single ☐ Divorced/separated ☐ Widowed

Do you have children? ☐ YES ☐ NO

Are you in paid work? ☐ YES, full time ☐ YES, part time ☐ NO

If YES, what is your main occupation: _____

If NOT in paid work, are you at present:

- | | | |
|--|--|--|
| <input type="checkbox"/> unemployed, out of work | <input type="checkbox"/> full time student | <input type="checkbox"/> housewife/husband |
| <input type="checkbox"/> retired | <input type="checkbox"/> disabled or too ill to work | |

If not currently employed, what is your usual (or last) occupation:

Do you have a car? ☐ NO ☐ YES, one ☐ YES, more than one

Is your accommodation: ☐ owned by you or your family ☐ rented

1. How do you pass urine?

- Without any assistance ☐
- Regular intermittent self catheterisation ☐
- Permanent/indwelling urinary catheter ☐
- Urinary stoma/ileal conduit ☐
- Mitroffanof ☐

When answering the following questions, please think about how you have been, on average, over the past four weeks.

2a. Do you often leak urine? (Tick one box)

- Never ☐
- About once per week or less often ☐
- 2 or 3 times a week ☐
- About once a day ☐
- Several times a day ☐
- All the time ☐

2b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a great deal)

0 1 2 3 4 5 6 7 8 9 10

3. When does the urine leak? (Please tick all that apply to you)

- Never – urine does not leak ☐
- Leaks before you can get to the toilet ☐
- Leaks when you cough or sneeze ☐
- Leaks when you are asleep ☐
- Leaks when you are physically active/exercising ☐
- Leaks when you have finishes urinating and are dressed ☐
- Leaks for no obvious reason ☐
- Leaks all the time ☐

Sometimes people try to protect themselves against urine loss by wearing pads, tissue paper or other protection.

4a. In the past four weeks have you used any protection? (Tick one box)

- Never ☐
- Some of the time ☐
- Most of the time ☐
- All of the time ☐

Go to Q 5

4b. If you did use protection in the past four weeks, what kind of protection did you use? (Tick as many as apply)

- Tissue/toilet paper/cloth ☐
- Minipads/pantliners ☐
- Sanitary/incontinence/other pads ☐
- Something else – please describe what you use ☐

4c. How many times each day did you change your protection?

- Never ☐
- 1-2 times ☐
- 3-5 times ☐
- 6 or more times ☐

We would like to know how much urine you think leaks

5a. How much urine do you usually leak (whether you wear protection or not)?

- None ☐
- A small amount ☐
- A moderate amount ☐
- A large amount ☐

5b. How much was the worst leakage over the past four weeks?

- None ☐
- A small amount ☐
- A moderate amount ☐
- A large amount ☐

PART 2 - EVERYDAY LIFE

Please think about the symptoms you may have had over the past 4 weeks.

6. How much does your urinary leakage affect your household jobs (for example, housework, DIY)?

- Not at all ☐
- A little ☐
- Moderately ☐
- A lot ☐

7. How much does your urinary leakage affect your life outside the house (e.g. shopping, visiting friends)?

- Not at all ☐
- A little ☐
- Moderately ☐
- A lot ☐

8. How much does your urinary leakage affect your job/employment?

- Not applicable to me ☐
- Not at all ☐
- A little ☐
- Moderately ☐
- A lot ☐

9. How much does your urinary leakage affect you ability to be actives (e.g. walking, doing exercise, playing with children)?

- Not at all ☐
- A little ☐
- Moderately ☐
- A lot ☐

10. Do you worry about where the toilets are when you go to a place you are not familiar with?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

11. Do you cut down on the amount you drink to reduce your urinary leakage?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

12. Do you avoid travelling because of your urinary leakage (e.g. by car, bus, train)?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

13. How much do you feel that your social life has been spoilt by urinary leakage during the past four weeks?

Please ring a number between 0 (not at all) and 10 (a great deal)

0 1 2 3 4 5 6 7 8 9 10

14. Overall, how much does your urinary leakage interfere with you everyday life?

Please ring a number between 0 (not at all) and 10 (a great deal)

0 1 2 3 4 5 6 7 8 9 10

15. In general, how would you rate the overall quality of your life during the past 4 weeks?

Please ring a number between 0 (worst quality of life) and 10 (best quality of life)

0 1 2 3 4 5 6 7 8 9 10

PART 3 - SEXUAL MATTERS

Please think about symptoms you may have had over the past 4 weeks.

16. Do you have vaginal pain or discomfort?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

17. Do you have a sex life at present?

- Yes ☐
No, because of my urinary leakage ☐
No, because of other reasons ☐

If NO please go to question 20

18. Do you have pain when you have sexual intercourse?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

19. Do you leak urine when you have sexual intercourse?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

20. How much do you feel that your sex life has been spoilt by urinary leakage during the past four weeks?

Please ring a number between 0 (not at all) and 10 (a great deal)

0 1 2 3 4 5 6 7 8 9 10

Not applicable to me ☐

PART 4 -EMOTIONAL ASPECTS

Please think about how you have been feeling over the past 4 weeks.

21. Does your urinary leakage make you feel depressed?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

22. Does your urinary leakage make you feel anxious or nervous?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

23. Does your urinary leakage make you feel frustrated?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

24. Do you ever feel embarrassed because of your urinary leakage?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

25. Does your urinary leakage reduce your enjoyment of life?

- Not at all ☐
A little ☐
Moderately ☐
A lot ☐

PART 5 - OTHER URINARY SYMPTOMS

We would like to know about any other symptoms you may have experienced over the past 4 weeks.

26a. How often do you pass urine during the day?

- Hourly ☐
Every 2 hours ☐
Every 3 hours ☐
Every 4 hours or more ☐

26b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

27a. During the night, how many times do you have to get up to urinate, on average?

- None ☐
One ☐
Two ☐
Three ☐
Four or more ☐

27b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

28a. Do you have to rush to the toilet to urinate?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

28b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

29a. Do you have pain in your bladder?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

29b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

30a. Is there a delay before you can start to urinate?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

30b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

31a. Do you have to strain to continue urinating?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

31b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

32a. Do you stop and start more than once while you urinate?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

32b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

33a. Would you say that the strength of your urinary stream is

- Normal ☐
Occasionally reduced ☐
Sometimes reduced ☐
Reduced most of the time ☐
Reduced all of the time ☐

33b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

34a. Do you have a burning feeling when you urinate?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

34b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

35a. How often do you feel that your bladder has not emptied properly after you have urinated?

- Never ☐
Occasionally (less than one third of the time) ☐
Sometimes (Between one and two thirds of the time) ☐
Most of the time (more than two thirds of the time) ☐
All of the time ☐

35b. How much of a problem is this for you?

Please ring a number between 0 (not at all) and 10 (a serious problem)

0 1 2 3 4 5 6 7 8 9 10

36. Overall, how much do your urinary symptoms interfere with your life?

Please ring a number between 0 (not at all) and 10 (a great deal)

0 1 2 3 4 5 6 7 8 9 10

PART 6 BOWEL FUNCTION

1. How do you go to the toilet?

- Without any assistance ☐
Regular laxatives ☐
Regular suppositories ☐
Regular rectal enemas ☐
Regular antegrade enemas (via a small opening in your tummy) ☐
I have a stoma ☐

If you do not have a stoma then please go to Part 7.

2. If you have a stoma, how long have you had this (approximately)?

3. Is the stoma permanent (not for reversal in the near future)?

YES / NO

4. Do you know what type of stoma you have?

Please could you give details below, and then please go to Part 9

PART 7 CONSTIPATION SCORE

Please answer the following questions based upon your experiences over the last 4 weeks.

1. How frequent are your bowel movements?

- 1-2 times per 1-2 days ☐
2 times per week ☐
Once per week ☐
Less than once per week ☐
Less than once per month ☐

2. Do you have any difficulty or pain on evacuation effort?

- Never ☐
Rarely ☐
Sometimes ☐
Usually ☐
Always ☐

3. Completeness: feeling incomplete evacuation

- Never ☐
Rarely ☐
Sometimes ☐
Usually ☐
Always ☐

4. Do you suffer from abdominal (tummy) pain?

- Never ☐
Rarely ☐
Sometimes ☐
Usually ☐
Always ☐

5. How long, on average do you spend in the lavatory per attempt?

- Less than 5 minutes ☐
5-10 minutes ☐
10-20 minutes ☐
20-30 minutes ☐
More than 30 minutes ☐

6. Do you require assistance to open your bowels?

- No assistance required ☐
Simulative laxatives ☐
Digital assistance or enemas ☐

7. How many times in 24 hours do you try to evacuate unsuccessfully (without passing faeces)?

- Never ☐
1 – 3 ☐
3 – 6 ☐
6 – 9 ☐
More than 9 ☐

8. How long have you been constipated for?

- Less than a year ☐
1 – 5 years ☐
5 – 10 years ☐
10 – 20 years ☐
More than 20 years ☐

PART 8 INCONTINENCE SCORE

Please tick the appropriate box for the symptoms you have experienced over the last 4 weeks.

Symptom	Never	Rarely	Sometimes	Weekly	Daily
Incontinence for solid stool					
Incontinence for liquid stool					
Incontinence for flatus (wind)					
Alteration to social life					

Do you need to wear a pad or plug?

YES / NO

Do you take constipation medications?

YES / NO

Are you able to defer defaecation for 15 minutes?

YES / NO

PART 9 OVERALL HEALTH

The following questions ask for your views about your health and how you feel about life in general. Do not spend too much time answering, as your immediate response is likely to be the most accurate.

1. In general, would you say your health is:

- Excellent ☐
 Very good ☐
 Good ☐
 Fair ☐
 Poor ☐

2. Compared to 3 months ago, how would you rate your health in general now?

- Much better than 3 months ago ☐
 Somewhat better than 3 months ago ☐
 About the same ☐
 Somewhat worse now than 3 months ago ☐
 Much worse now than 3 months ago ☐

3. The following questions are about activities you might do during a typical day. Does your health limit you in these activities? If so, how much?

(Please tick one box on each line)

Activities	Yes, limited a lot	Yes, limited a little	No, not limited at all
a) Vigorous activities , such as running, lifting heavy objects, participating in strenuous sports			
b) Moderate activities , such as moving a table, pushing a vacuum, bowling or playing golf			
c) Lifting or carrying groceries			
d) Climbing several flights of stairs			
e) Climbing one flight of stairs			
f) Bending kneeling or stooping			
g) Walking more than a mile			
h) Walking half a mile			
i) Walking 100 yards			
j) Bathing and dressing yourself			

4. During the past 2 weeks, have you had any of the following problems with your work or other regular daily activities as a result of your physical health? (Please tick one box on each line)

	YES	NO
a) Cut down on the amount of time you spent on work or other activities		
b) Accomplished less than you would like		
c) Were limited in the kind of work or other activities		
d) Had difficulty performing the work or other activities e.g. it took more effort		

5. During the past 2 weeks, how much time have you had any of the following problems with your work or other regular daily activities as a result of any emotional problems (such as feeling depressed or anxious)? (Please tick one box) on each line

	YES	NO
a) Cut down on the amount of time you spent on work or other activities		
b) Accomplished less than you would like		
c) Didn't do work or other activities as carefully as usual		

6. During the past 2 weeks, to what extent have your physical health or emotional problems interfered with your normal social activities with family, neighbours or groups?

- Not at all ☐
 Slightly ☐
 Moderately ☐
 Quite a bit ☐
 Extremely ☐

7. How much bodily pain have you had during the past 2 weeks ?

- None ☐
 Very mild ☐
 Mild ☐
 Moderate ☐
 Severe ☐
 Very Severe ☐

8. During the past 2 weeks, how much did pain interfere with your normal work (including both outside the home and housework)?

- Not at all ☐
 Slightly ☐
 Moderately ☐
 Quite a bit ☐
 Extremely ☐

9. For each question please give one answer that comes closest to the way you have been feeling. How much time during the last 2 weeks:

	All of the time	Most of the time	A good bit of the time	Some of the time	A little of the time	None of the time
a) Did you feel full of life?						
b) Have you been a very nervous person?						
c) Have you felt so down in the dumps that nothing would cheer you up?						
d) Have you felt calm and peaceful?						
e) Did you have a lot of energy?						
f) Have you felt downhearted and low?						
g) Did you feel worn out?						
h) Have you been a happy person?						
i) Did you feel tired?						

10. During the past 2 weeks, how much of the time has your physical health or emotional problems interfered with your social activities (like visiting friends, relatives etc.).

- All of the time ☐
 Most of the time ☐
 Some of the time ☐
 A little of the time ☐
 None of the time ☐

11. How TRUE or FALSE is each of the following statements for you?
(Please tick one box)

	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
a) I seem to get ill more easily than other people					
b) I am as healthy as anybody I know					
c) I expect my health to get worse					
d) My health is excellent					

PART 10 - GYNAECOLOGY

The following questions relate to menstruation (periods).

1. Do you have regular periods? YES / NO
If YES then please answer questions 2 – 7
If NO then go to question 8
2. Did you require any operations to help you have periods? YES / NO
3. What was the age of menarche (1st period)
4. Do you have a regular cycle? YES / NO
5. Are your periods painful? YES / NO
6. Do you have problems with heavy bleeding? YES / NO
7. Are you able to use tampons? YES / NO
8. Have you had a hysterectomy (womb removed)? YES / NO
9. Are you currently taking hormone tablets? YES / NO
10. Are you being investigated for amenorrhoea (no periods)? YES / NO

The following questions relate to sexual activity

1. Are you sexually active? YES / NO
2. At what age did you become sexually active?
3. Did you require any dilator treatment prior to sexual intercourse? YES / NO
4. Did you require any operations to help you become sexually active? YES / NO
5. Do you experience any difficulty in penetration? YES / NO

PART 11 - OBSTETRIC HISTORY

1. Have you been or are you currently pregnant? YES / NO

If yes please give details of all pregnancies and outcome, please include details of any miscarriages and/or terminations if applicable.

Pregnancies ending before 24 weeks

Date	Outcome	Gestational stage	Problems related to anorectal malformation	Problems related to pregnancy

Pregnancies ending after 24 weeks

Date	Outcome	Gestational Age	Problems related to anorectal malformation	Problems related to pregnancy

Pregnancy outcome may be one of the following:

Spontaneous vaginal

Induced vaginal

Forceps Vaginal

Ventouse Vaginal

Elective Caesarean

Emergency Caesarean

Did you require any fertility assistance with any or all of the above pregnancies? If so please give details.

PART 12 – SEXUALITY

Listed below are several statements that concern the topic of sexual relationships. Please read each item carefully and decide to what extent it is characteristic of you. Whenever possible, answer the questions with your most recent partner in mind. If you have never had a sexual relationship, answer in terms of what you think your responses would most likely be. Then, for each statement fill in the response on the answer sheet that indicates how much it applies to you by using the following scale:

- A = Not at all characteristic of me.**
B = Slightly characteristic of me.
C = Somewhat characteristic of me.
D = Moderately characteristic of me.
E = Very characteristic of me.

Statement	Response
1. I am confident about myself as a sexual partner.	
2. I feel anxious when I think about the sexual aspects of my life.	
3. I'm very assertive about the sexual aspects of my life.	
4. I am depressed about the sexual aspects of my life.	
5. I am somewhat afraid of becoming sexually involved with another person.	
6. I am very satisfied with the way my sexual needs are currently being met.	
7. I am a pretty good sexual partner.	
8. I'm worried about the sexual aspects of my life.	
9. I'm not very direct about voicing my sexual preferences.	
10. I am disappointed about the quality of my sex life.	
11. I sometimes have a fear of sexual relationships.	
12. I am very satisfied with my sexual relationship.	
13. I am better at sex than most other people.	
14. Thinking about the sexual aspects of my life leaves me with an uneasy feeling.	
15. I am somewhat passive about expressing my sexual desires.	
16. I feel discouraged about my sex life.	
17. I sometimes am fearful of sexual activity.	
18. My sexual relationship meets my original expectations.	
19. I would rate myself pretty favourably as a sexual partner.	
20. I usually worry about the sexual aspects of my life.	
21. I do not hesitate to ask for what I want in a sexual relationship.	
22. I feel unhappy about my sexual relationships.	
23. I don't have very much fear about engaging in sex.	
24. My sexual relationship is very good compared to most.	
25. I would be very confident in a sexual encounter.	
26. I feel nervous when I think about the sexual aspects of my life.	
27. When it comes to sex, I usually ask for what I want.	
28. I feel sad when I think about my sexual experiences.	

29. I'm not very afraid of becoming sexually active.	
30. I am very satisfied with the sexual aspects of my life.	
31. I responded to the above items based on: A) A current sexual relationship. B) A past sexual relationship. C) An imagined sexual relationship.	

PART 13 - BODY ESTEEM

Instructions

On this page are listed a number of body parts and functions. Please read each item and indicate how you feel about this part or function of your own body using the following scale:

- 1 = Have strong negative feelings**
- 2 = Have moderate negative feelings**
- 3 = Have no feeling one way or the other**
- 4 = Have moderate positive feelings**
- 5 = Have strong positive feelings**

BODY PART	SCORE
Body scent	
Appetite	
Nose	
Physical stamina	
Reflexes	
Lips	
Muscular strength	
Waist	
Energy level	
Thighs	
Ears	
Biceps	
Chin	
Body build	
Physical coordination	
Buttocks	
Agility	
Width of shoulders	
Arms	
Chest or breasts	
Appearance of eyes	
Cheeks/cheekbones	
Hips	
Legs	
Figure/physique	
Sex drive	
Feet	
Sex organs	
Appearance of stomach	
Health	
Sex activities	
Body hair	
Physical condition	
Face	
Weight	

Appendix 4

Survey into Adult Outcomes after
Anorectal Malformation and Cloaca
Repair.



CONFIDENTIAL

CONSENT FORM

Survey into Adult Outcomes after Anorectal Malformation and Cloaca Repair.

Please read the information sheet.

Thank you for agreeing to take part in our study, please could you complete the questionnaires and return them in the stamped addressed envelope enclosed. If you have any queries relating to the study or and difficulties with the questionnaires please contact Dr Melissa Davies on the details above.

Please read and delete as appropriate:

Have you read the information about this study ? YES/NO

Have you received enough information about this study? YES/NO

Do you understand that you are free to withdraw from this study

- at any time?
- without having to give a reason?
- without affecting your future medical care? YES/NO

Do you agree to take part in this study? YES/NO

Name of patient

Date

Signature

If you have any comments or concerns you may discuss these with the investigator. If you wish to go further and complain about any aspect of the way you have been approached or treated during the course of the study, you should write or get in touch with the Complaints Manager, UCL hospitals. Please quote the UCLH project number at the top this consent form.

University College London
86-96 Chenies Mews
London WC1E 6HX

Tel: 020 7679 6654
Fax: 020 7383 7429

Head of Department Professor CH Rodeck DSc, FRCOG, FRCPath



Access to Medical Notes

We would like to be able to view your medical records from other hospitals where you have received treatment. This is gain a complete record of all operations, investigations and treatment you have had.

To do this we would like your permission to be able to look at your notes from other hospitals where you have been a patient.

If you agree to this please complete the form below.

Full Name (current)

Previous names used

Date of Birth

Current Address

.....

Hospitals where you have been treated

.....

.....

.....

.....

I give permission for Dr Melissa Davies of University College Lodon to view my hospital case notes

Signature.....

Date.....

Social and Demographic Information

This section tells us whether our survey sample is similar to the UK population in social and economic terms. The information is not for identifying you.

Your age:

Your gender: Male / Female

Which best describes your ethnic background?

- | | | | |
|--------------------------------------|--|--|--|
| <input type="checkbox"/> Bangladeshi | <input type="checkbox"/> Black African | <input type="checkbox"/> Black British | <input type="checkbox"/> Black Caribbean |
| <input type="checkbox"/> Black other | <input type="checkbox"/> British Asian | <input type="checkbox"/> Chinese | <input type="checkbox"/> Indian |
| <input type="checkbox"/> Pakistani | <input type="checkbox"/> White | <input type="checkbox"/> | |
- Other: _____

What is the highest qualification that you have passed?

- | | |
|--|---|
| <input type="checkbox"/> Degree or equivalent | <input type="checkbox"/> Non-degree teaching or other qualification |
| <input type="checkbox"/> 'A' level or equivalent | <input type="checkbox"/> GCSE, 'O' level or equivalent |
| <input type="checkbox"/> GCSE or equivalent | <input type="checkbox"/> Other qualifications <input type="checkbox"/> No qualification |

Are you currently in a relationship? (Tick one box only of six)

- YES: ☐ Married, living together ☐ Not married, living together ☐ Not living together
- NO: ☐ Single ☐ Divorced/separated ☐ Widowed

Do you have children? ☐ YES ☐ NO

Are you in paid work? ☐ YES, full time ☐ YES, part time ☐ NO

If YES, what is your main occupation: _____

If NOT in paid work, are you at present:

- | | | |
|--|--|--|
| <input type="checkbox"/> unemployed, out of work | <input type="checkbox"/> full time student | <input type="checkbox"/> housewife/husband |
| <input type="checkbox"/> retired | <input type="checkbox"/> disabled or too ill to work | |

If not currently employed, what is your usual (or last) occupation:

Do you have a car? ☐ NO ☐ YES, one ☐ YES, more than one

Is your accommodation: ☐ owned by you or your family ☐ rented

PART 1 – URINARY SYMPTOMS

How do you pass urine?

- without any assistance ☐
- regular intermittent self catheterisation ☐
- permanent/indwelling urinary catheter ☐
- urinary stoma/ileal conduit ☐
- Mitroffanoff ☐

Please answer both parts of each question, thinking about the **symptoms you have experienced in the last month**.

You will see that some questions ask if you have a symptom occasionally, sometimes or most of the time.

Occasionally	=	less than one third of the time
Sometimes	=	between one and two thirds of the time
Most of the time	=	more than two thirds of the time

1. During the day, how many times do you urinate, on average?

- 1 to 6 times ☐
- 7 to 8 times ☐
- 9 to 10 times ☐
- 11 to 12 times ☐
- 13 times or more ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

2. During the night, how many times do you have to get up to urinate on average?

- none ☐
- one ☐
- two ☐
- three ☐
- four or more ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

3. Do you have to rush to the toilet to urinate?

- never ☐
- occasionally ☐

- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

4. Does urine leak before you can get to the toilet?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

5. Do you have pain in your bladder?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

6. Does urine leak when you cough or sneeze?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

7. Do you ever leak for no obvious reason and without feeling that you want to go?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

8. Is there a delay before you can start to urinate?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

9. Do you have to strain to start urinating?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

10. Do you have to strain to continue urinating?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

11. Do you usually urinate standing up or sitting down?

- Standing up ☐
Sitting down ☐

How much of a problem is this for you?

- not a problem ☐
a bit of a problem ☐
quite a problem ☐
a serious problem ☐

12. Would you say that the strength of your urinary stream is...

- normal ☐
occasionally reduced ☐
sometimes reduced ☐
reduced most of the time ☐
reduced all of the time ☐

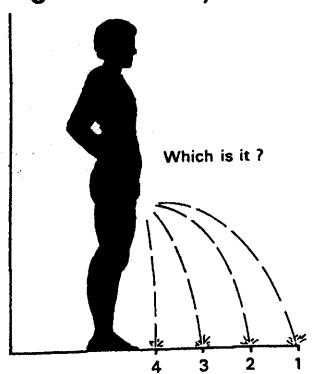
How much of a problem is this for you?

- not a problem ☐
a bit of a problem ☐
quite a problem ☐
a serious problem ☐

13. Do you think you have always had a weak stream?

- Yes ☐
No ☐

14 Would you say that the strength of your urinary stream is... (please ring one number)



(from Peeling, 1989)

15. Do you stop and start more than once while you urinate?

- never ☐
occasionally ☐
sometimes ☐

How much of a problem is this for you?

- most of the time ☐
- all of the time ☐

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

16. Do you have a burning feeling when you urinate?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

17. How often do you feel that your bladder has not emptied properly after you have urinated?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

18. Does your stream end with a dribble?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

19. How often have you had a slight wetting of your pants a few minutes after you had finished urinating?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

20. Do you ever leak urine when you are asleep?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

21. If you leak urine during the day, do you have to change your clothes or wear pads?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

22. Do you have to urinate again (within 15 minutes) after you thought you had finished urinating?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

23. Have you ever blocked up completely so that you could not urinate at all and had to have a catheter passed to drain the bladder?

- No ☐
- Yes, once ☐
- Yes, twice ☐
- Yes, more than twice ☐

24. To what extent do you feel that your sex life has been spoilt by your urinary symptoms?

- not at all ☐
- a little ☐
- somewhat ☐
- a lot ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

25. Do you get erections?

- yes, with normal rigidity ☐
- yes, with reduced rigidity ☐
- yes, with severely reduced rigidity ☐
- no, erection not possible ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

26. Do you have an ejaculation of semen?

- yes, normal quantity ☐
- yes, reduced quantity ☐
- yes, significantly reduced quantity ☐
- no ejaculation ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

27. Do you have pain or discomfort during ejaculation?

- no ☐
- yes, slight pain/discomfort ☐
- yes, moderate pain/discomfort ☐
- yes, severe pain/discomfort ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

28. How often do you pass urine during the day?

- hourly ☐
- every 2 hours ☐
- every 3 hours ☐
- every 4 hours ☐

How much of a problem is this for you?

- not a problem ☐
- a bit of a problem ☐
- quite a problem ☐
- a serious problem ☐

29. Do you cut down on the amount you drink so that your urinary symptoms improve and you can do the things you want to do?

- never ☐
- occasionally ☐
- sometimes ☐
- most of the time ☐
- all of the time ☐

30. Overall, how much do your urinary symptoms interfere with your life?

- not at all ☐
- a little ☐
- somewhat ☐
- a lot ☐

31. How long have you had urinary symptoms that bother you?

- less than one year – give months ☐
- between one year and two years ☐
- between two and three years ☐
- more than three years ☐

32. Do you have any worries about your urinary problems?

Please list below:

33. If you had to spend the rest of your life with your urinary symptoms as they are now, how would you feel?

- perfectly happy ☐
- pleased ☐
- mostly satisfied ☐
- mixed feelings ☐
- mostly dissatisfied ☐
- very unhappy ☐
- desperate ☐

34. Which of your urinary symptoms bother you the most at the moment?

Please list the symptoms that bother you the most below.

- 1.
- 2.
- 3.

PART 2 - BOWEL FUNCTION

1. How do you go to the toilet?

- Without any assistance ☐
Regular laxatives ☐
Regular suppositories ☐
Regular rectal enemas ☐
Regular antegrade enemas (via a small opening in your tummy) ☐
I have a stoma ☐

If you do not have a stoma then please go to Part 3

2. If you have a stoma, how long have you had this (approximately)?

3. Is the stoma permanent (not for reversal in the near future)?

YES / NO

4. Do you know what type of stoma you have?

Please could you give details below.

PART 3 - CONSTIPATION SCORE

Please answer the following questions based upon your experiences over the last 4 weeks.

1. How frequent are your bowel movements?

- 1-2 times per 1-2 days ☐
2 times per week ☐
Once per week ☐
Less than once per week ☐
Less than once per month ☐

2. Do you have any difficulty or pain on evacuation effort?

- Never ☐
Rarely ☐
Sometimes ☐
Usually ☐
Always ☐

3. Completeness: feeling incomplete evacuation

- Never ☐
Rarely ☐
Sometimes ☐
Usually ☐
Always ☐

4. Do you suffer from abdominal (tummy) pain?

- Never ☐
- Rarely ☐
- Sometimes ☐
- Usually ☐
- Always ☐

5. How long, on average do you spend in the lavatory per attempt?

- Less than 5 minutes ☐
- 5-10 minutes ☐
- 10-20 minutes ☐
- 20-30 minutes ☐
- More than 30 minutes ☐

6. Do you require assistance to open your bowels?

- No assistance required ☐
- Simulative laxatives ☐
- Digital assistance or enemas ☐

7. How many times in 24 hours do you try to evacuate unsuccessfully (without passing faeces)?

- Never ☐
- 1 – 3 ☐
- 3 – 6 ☐
- 6 – 9 ☐
- More than 9 ☐

8. How long have you been constipated for?

- Less than a year ☐
- 1 – 5 years ☐
- 5 – 10 years ☐
- 10 – 20 years ☐
- More than 20 years ☐

PART 4 - INCONTINENCE SCORE

Please tick the appropriate box for the symptoms you have experienced **over the last 4 weeks.**

Symptom	Never	Rarely	Sometimes	Weekly	Daily
Incontinence for solid stool					
Incontinence for liquid stool					
Incontinence for flatus (wind)					
Alteration to social life					

Do you need to wear a pad or plug?

YES / NO

Do you take constipation medications?

YES / NO

Are you able to defer defaecation for 15 minutes?

YES / NO

PART 5 - OVERALL HEALTH

The following questions ask for your views about your health and how you feel about life in general. Do not spend too much time answering, as your immediate response is likely to be the most accurate.

1. In general, would you say your health is:

- Excellent ☐
 Very good ☐
 Good ☐
 Fair ☐
 Poor ☐

2. Compared to 3 months ago, how would you rate your health in general now?

- Much better than 3 months ago ☐
 Somewhat better than 3 months ago ☐
 About the same ☐
 Somewhat worse now than 3 months ago ☐
 Much worse now than 3 months ago ☐

3. The following questions are about activities you might do during a typical day. Does your health limit you in these activities? If so, how much?
 (Please tick **one** box on each line)

Activities	Yes, limited a lot	Yes, limited a little	No, not limited at all
a) Vigorous activities , such as running, lifting heavy objects, participating in strenuous sports			
b) Moderate activities , such as moving a table, pushing a vacuum, bowling or playing golf			
c) Lifting or carrying groceries			
d) Climbing several flights of stairs			
e) Climbing one flight of stairs			
f) Bending kneeling or stooping			
g) Walking more than a mile			
h) Walking half a mile			
i) Walking 100 yards			
j) Bathing and dressing yourself			

4. During the past 2 weeks, have you had any of the following problems with your work or other regular daily activities as a result of your physical health? (Please tick **one** box on each line)

	YES	NO
a) Cut down on the amount of time you spent on work or other activities		
b) Accomplished less than you would like		
c) Were limited in the kind of work or other activities		
d) Had difficulty performing the work or other activities e.g. it took more effort		

5. During the past 2 weeks, how much time have you had any of the following problems with your work or other regular daily activities as a result of any emotional problems (such as feeling depressed or anxious)?
(Please tick **one** box) on each line

	YES	NO
a) Cut down on the amount of time you spent on work or other activities		
b) Accomplished less than you would like		
c) Didn't do work or other activities as carefully as usual		

6. During the past 2 weeks, to what extent have your physical health or emotional problems interfered with your normal social activities with family, neighbours or groups?

- Not at all ☐
 Slightly ☐
 Moderately ☐
 Quite a bit ☐
 Extremely ☐

7. How much bodily pain have you had during the past 2 weeks ?

- None ☐
 Very mild ☐
 Mild ☐
 Moderate ☐
 Severe ☐
 Very Severe ☐

8. During the past 2 weeks, how much did pain interfere with your normal work (including both outside the home and housework)?

- Not at all ☐
 Slightly ☐
 Moderately ☐
 Quite a bit ☐
 Extremely ☐

9. For each question please give one answer that comes closest to the way you have been feeling. How much time during the last 2 weeks:

	All of the time	Most of the time	A good bit of the time	Some of the time	A little of the time	None of the time
a) Did you feel full of life?						
b) Have you been a very nervous person?						
c) Have you felt so down in the dumps that nothing would cheer you up?						
d) Have you felt calm and peaceful?						
e) Did you have a lot of energy?						
f) Have you felt downhearted and low?						
g) Did you feel worn out?						
h) Have you been a happy person?						
i) Did you feel tired?						

10. During the past 2 weeks, how much of the time has your physical health or emotional problems interfered with your social activities (like visiting friends, relatives etc.).

- All of the time ☐
 Most of the time ☐
 Some of the time ☐
 A little of the time ☐
 None of the time ☐

11. How TRUE or FALSE is each of the following statements for you?
 (Please tick **one** box)

	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
a) I seem to get ill more easily than other people					
b) I am as healthy as anybody I know					
c) I expect my health to get worse					
d) My health is excellent					

PART 6 – MALE FERTILITY & SEXUAL FUNCTION

Are you sexually active? YES / NO

At what age did you first become sexually active?

Have you fathered any children? YES / NO

If **YES** please give details of dates of deliveries (please include miscarriages and terminations).

Date	Outcome

Did you require any fertility assistance e.g IVF?

YES / NO

If YES please give details.

SEXUAL DRIVE

If we define sexual drive as a feeling that may include wanting to have a sexual experience (masturbation or intercourse), thinking about having sex, or feeling frustrated due to the lack of sex.

1. During the last 30 days, how many days have you felt sexual drive?

- No days ☐
- Only a few days ☐
- Some days ☐
- Most days ☐
- Almost every day ☐

2. During the past 30 days, how would you rate the level of your sexual drive?

- None at all ☐
- Low ☐
- Medium ☐
- Medium high ☐
- High ☐

ERECTIONS

3. Over the past 30 days, how often have you had partial or full sexual erections when you were sexually stimulated in any way?

- Not at all ☐
- A few times ☐
- Fairly often ☐
- Usually ☐
- Always ☐

4. Over the past 30 days, when you had erections, how often were they firm enough to have sexual intercourse?

- 0 ☐
- 1 ☐
- 2 ☐
- 3 ☐
- 4 ☐

5. How much difficulty did you have getting an erection during the past 30 days?

- Did not get erections at all ☐
- A lot of difficulty ☐
- Some difficulty ☐
- Little difficulty ☐
- No difficulty ☐

EJACULATION

6. In the past 30 days, how much difficulty have you had ejaculating when you have been sexually stimulated?

- No sexual stimulation in last 30 days ☐
- A lot of difficulty ☐
- Some difficulty ☐
- Little difficulty ☐
- No difficulty ☐

7. In the past 30 days, how much did you consider the amount of semen you ejaculate to be a problem for you?

- Did not climax ☐
- Big problem ☐
- Medium problem ☐
- Small problem ☐
- No problem ☐

PROBLEM ASSESSMENT

8. In the past 30 days, to what extent have you considered a lack of sex drive to be a problem?

- Big problem ☐
- Medium problem ☐
- Small problem ☐
- Very small problem ☐
- No problem ☐

9. In the past 30 days, to what extent have you considered your ability to get and keep erections to be a problem?

- Big problem ☐
- Medium problem ☐
- Small problem ☐
- Very small problem ☐
- No problem ☐

10. In the past 30 days, to what extent have you considered your ejaculation to be a problem?

- Big problem ☐
- Medium problem ☐
- Small problem ☐
- Very small problem ☐
- No problem ☐

OVERALL SATISFACTION

11. Overall, during the past 30 days, how satisfied have you been with your sex life?

- Very dissatisfied ☐
- Mostly dissatisfied ☐
- Neutral ☐
- Mostly satisfied ☐
- Very satisfied ☐

PART 7 – SEXUALITY

Listed below are several statements that concern the topic of sexual relationships. Please read each item carefully and decide to what extent it is characteristic of you. Whenever possible, answer the questions with your most recent partner in mind. If you have never had a sexual relationship, answer in terms of what you think your responses would most likely be. Then, for each statement fill in the response on the answer sheet that indicates how much it applies to you by using the following scale:

A = Not at all characteristic of me.
B = Slightly characteristic of me.
C = Somewhat characteristic of me.
D = Moderately characteristic of me.
E = Very characteristic of me.

Statement	Response
1. I am confident about myself as a sexual partner.	
2. I feel anxious when I think about the sexual aspects of my life.	
3. I'm very assertive about the sexual aspects of my life.	
4. I am depressed about the sexual aspects of my life.	
5. I am somewhat afraid of becoming sexually involved with another person.	
6. I am very satisfied with the way my sexual needs are currently being met.	
7. I am a pretty good sexual partner.	
8. I'm worried about the sexual aspects of my life.	
9. I'm not very direct about voicing my sexual preferences.	
10. I am disappointed about the quality of my sex life.	
11. I sometimes have a fear of sexual relationships.	
12. I am very satisfied with my sexual relationship.	
13. I am better at sex than most other people.	
14. Thinking about the sexual aspects of my life leaves me with an uneasy feeling.	
15. I am somewhat passive about expressing my sexual desires.	
16. I feel discouraged about my sex life.	
17. I sometimes am fearful of sexual activity.	
18. My sexual relationship meets my original expectations.	
19. I would rate myself pretty favourably as a sexual partner.	
20. I usually worry about the sexual aspects of my life.	
21. I do not hesitate to ask for what I want in a sexual relationship.	
22. I feel unhappy about my sexual relationships.	
23. I don't have very much fear about engaging in sex.	
24. My sexual relationship is very good compared to most.	
25. I would be very confident in a sexual encounter.	
26. I feel nervous when I think about the sexual aspects of my life.	

27. When it comes to sex, I usually ask for what I want.	
28. I feel sad when I think about my sexual experiences.	
29. I'm not very afraid of becoming sexually active.	
30. I am very satisfied with the sexual aspects of my life.	
31. I responded to the above items based on: A) A current sexual relationship. B) A past sexual relationship. C) An imagined sexual relationship.	

PART 8 - BODY ESTEEM

Instructions

On this page are listed a number of body parts and functions. Please read each item and indicate how you feel about this part or function of your own body using the following scale:

- 1 = Have strong negative feelings**
- 2 = Have moderate negative feelings**
- 3 = Have no feeling one way or the other**
- 4 = Have moderate positive feelings**
- 5 = Have strong positive feelings**

BODY PART	SCORE
Body scent	
Appetite	
Nose	
Physical stamina	
Reflexes	
Lips	
Muscular strength	
Waist	
Energy level	
Thighs	
Ears	
Biceps	
Chin	
Body build	
Physical coordination	
Buttocks	
Agility	
Width of shoulders	
Arms	
Chest or breasts	
Appearance of eyes	
Cheeks/cheekbones	
Hips	
Legs	
Figure/physique	
Sex drive	
Feet	
Sex organs	
Appearance of stomach	
Health	
Sex activities	
Body hair	
Physical condition	
Face	
Weight	

